Pediatric urinary disorders

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Learning objectives:

- Recall normal anatomy
- Identified the common congenital anomalies
- How to detect this anomaly on radiological investigation
- Important steps in management

2

Hypospadias

Laurence S. Baskin

I. INTRODUCTION

A. *Hypospadias* is a congenital defect of the penis, resulting in incomplete development of the anterior urethra, corpora cavernosa, and prepuce (foreskin).

B. Clinically, the hypospadiac urethral meatus does not cause significant symptoms other than a urinary stream, which may be deflected downward.

C. Hypospadias is also associated with penile curvature and may result in infertility secondary to difficulty in semen delivery.

D. Hypospadias is not associated with an increased risk of urinary tract infection.

II. EMBRYOLOGY

A. At 1 month gestation the male and female genitalia are essentially indistinguishable.

B. Under the influence of testosterone, the male external genitalia become masculinized.

C. By the end of the first trimester (at approximately 16 to 18 weeks), the penile urethra and accompanying prepuce are completely formed.

D. Abnormalities in this development can lead to hypospadias and associated penile anomalies.

E. In hypospadias, incomplete development of the glandular urethra does not allow the preputial folds to fuse.

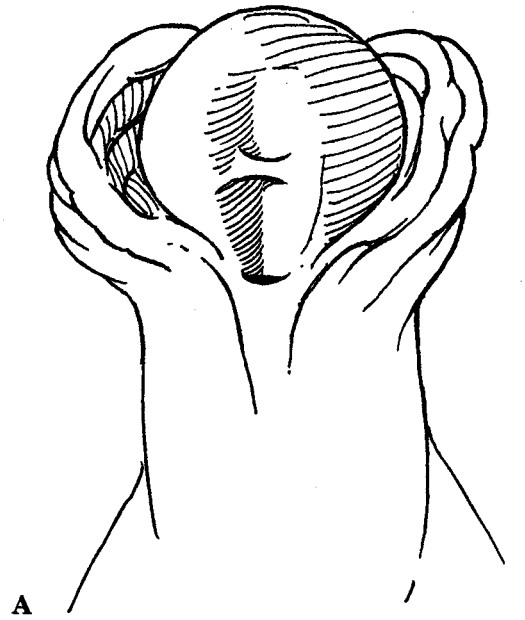
F. Consequently, in hypospadias the foreskin is absent on the ventrum and there is excessive foreskin on the dorsal surface (dorsal preputial hood).

III. CLASSIFICATION

A. Hypospadias can be classified as to the location of the urethral meatus without taking into account penile curvature. Penile curvature or chordee is germane in that a distal hypospadias with severe curvature will require extensive reconstruction to correct both the curvature and urethra, as the abnormal meatus, which may now be at the penoscrotal junction.

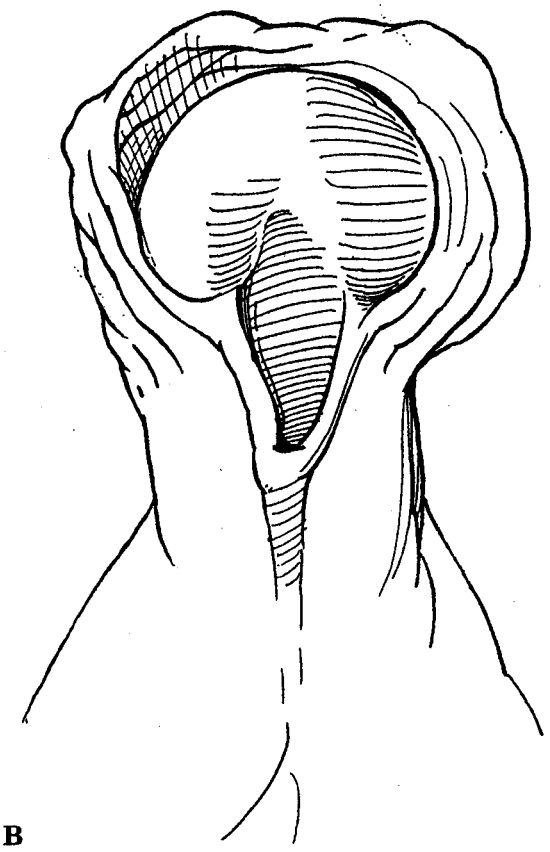
B. A more useful surgical classification is the location of the meatus after penile straightening or chordee correction at the time of reconstructive surgery, where

1. Fifty percent of patients have anterior hypospadias with the meatus on the glans or subcoronal (Fig. 2–1A).
2. Twenty percent have the urethral meatus on the penile shaft (Fig. 2–1B).
3. Thirty percent have the meatus between the perineum and the penoscrotal junction (Fig. 2–1C).



A

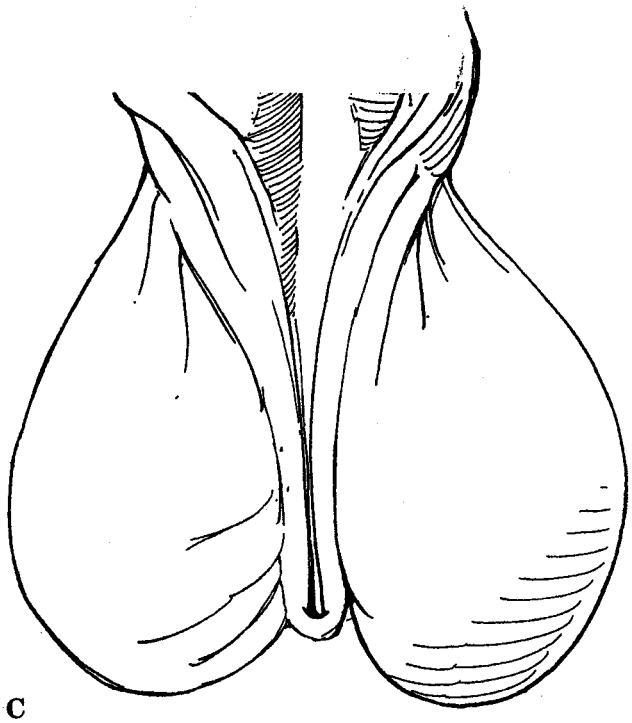
Mild



B

Moderate

Fig. 2-1. Classification of hypospadias. A: Anterior hypospadias. B: Penile shaft hypospadias.



C

Severe

Fig. 2–1. (*Continued*) **C: Scrotal hypospadias.**

IV. INCIDENCE, GENETICS, AND ETIOLOGY

A. Hypospadias occurs in 1 per 250 live male births.

B. There is a 14% incidence in male siblings and an 8% incidence in offspring.

C. The majority of cases of hypospadias have no known etiology. Extensive research into abnormal androgen metabolism or the levels of the androgen precursors, testosterone, or the more potent dihydrotestosterone has revealed only a small percentage of patients with any abnormalities.

D. Environmental pollutants or endocrine disruptors have been suggested to cause hypospadias by maternal exposure that is carried to the developing fetus. Case reports human data and experimental animal data are cause for concern and further investigation is warranted.

V. ASSOCIATED ANOMALIES

A. Undescended testes occur in approximately 9% of patients with hypospadias.

B. There is increased incidence of up to 30% in patients with penoscrotal or more severe hypospadias.

C. Inguinal hernias also occur in approximately 9% of patients with hypospadias.

D. A utricle or Müllerian remnant in the posterior urethra is found in a high percentage of patients with severe hypospadias.

E. Associated urinary tract anomalies are uncommon in patients with isolated hypospadias because the external genitalia are formed much later than the kidneys, ureter, and bladder.
F. Patients with hypospadias and an undescended testicle or an inguinal hernia do not need further urinary tract evaluation.
G. Patients who have hypospadias in association with other organ system anomalies such as a cardiac murmur, imperforate anus, limb malformations, cleft lip, or pyloric stenosis require renal and bladder imaging with an abdominal ultrasound.
H. Patients with severe hypospadias and undescended testes should be karyotyped and undergo a further endocrinologic workup (see Chapter 4).

VI. TREATMENT

A. There are five basic phases for the successful reconstruction of the hypospadiac penis:
   1. Creation of a normal urethral meatus and glans penis
   2. A straight penis
   3. A normal urethra
   4. Skin covering
   5. Normal position of the scrotum in relationship to the penis

VII. TIMING OF SURGERY

A. Hypospadias surgery is best performed between the ages of 6 and 18 months, prior to toilet teaching and during the psychological window when genital awareness has not been recognized by the patient.
B. Outpatient surgery is now the standard of care. The majority of hypospadiac defects can be repaired in a single-stage operation; severe cases often require a staged procedure. Early hypospadias repair with minimal hospitalization helps to avoid separation anxiety and fears related to genital surgery.

VIII. ANESTHESIA

A. Hypospadias surgery is performed under general anesthesia.
B. A penile nerve block or a caudal supplementation is standard to minimize postoperative discomfort.

IX. HYPOSPADIAS OPERATIONS

A. **Primary tubularization** (GAP, Thiersch-Duplay, King, Pyramid) (Fig. 2–2)
B. **Meatal advancement and glanuloplasty procedure** (MAGPI) (Fig. 2–3)
C. **Primary tubularization** with incision of the urethral plate (Snodgrass) (Fig. 2–4)
D. **Onlay island flap procedure** (Fig. 2–5)
E. **Staged repairs:** In patients with severe hypospadias, two planned operations performed about 6 months apart yield a controlled outcome with fewer complications. The first stage consists of penile straightening and the transfer of vascularized skin from the excess prepuce from the dorsal to the ventral

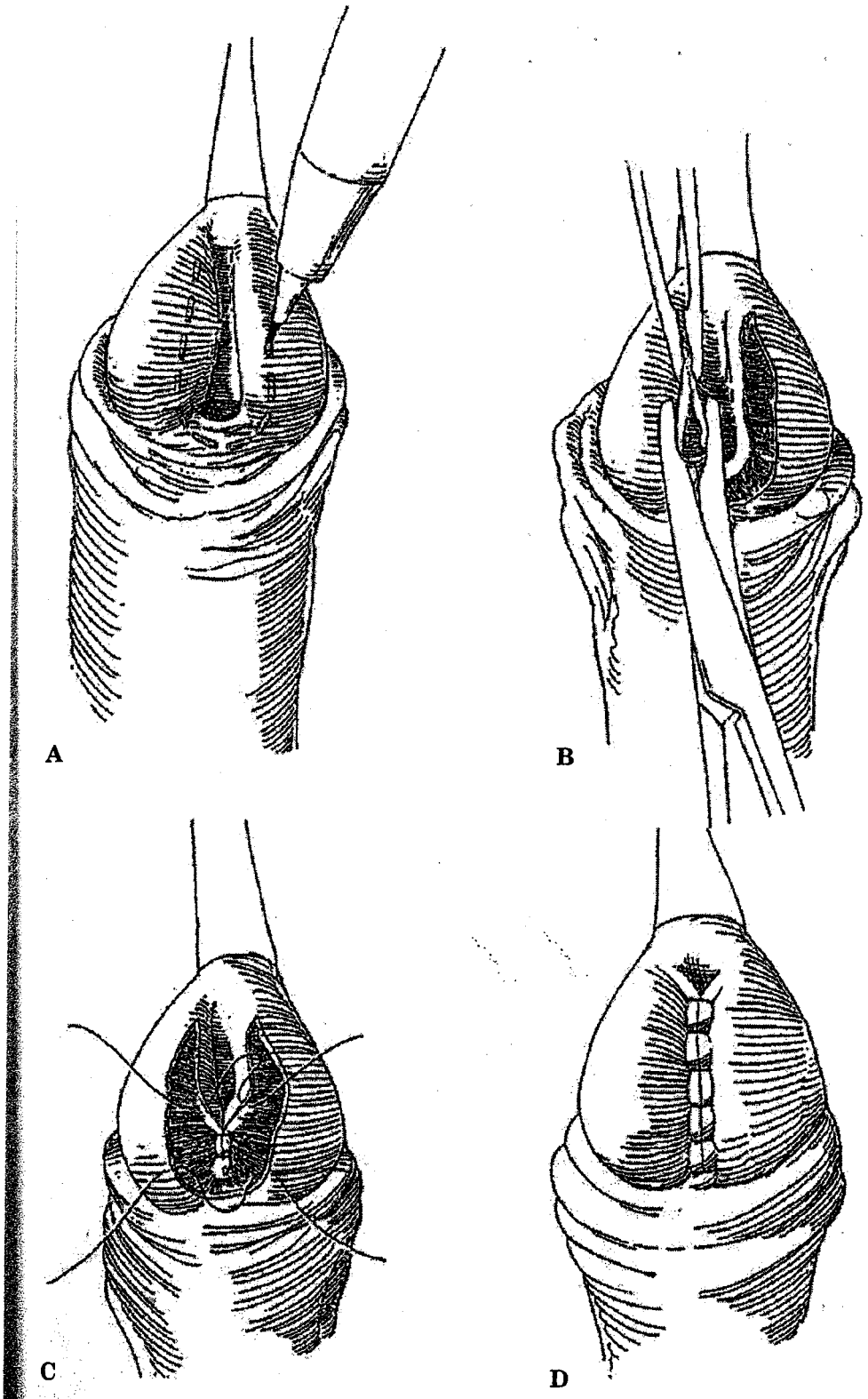


Fig. 2–2. A–D: GAP hypospadias repair.

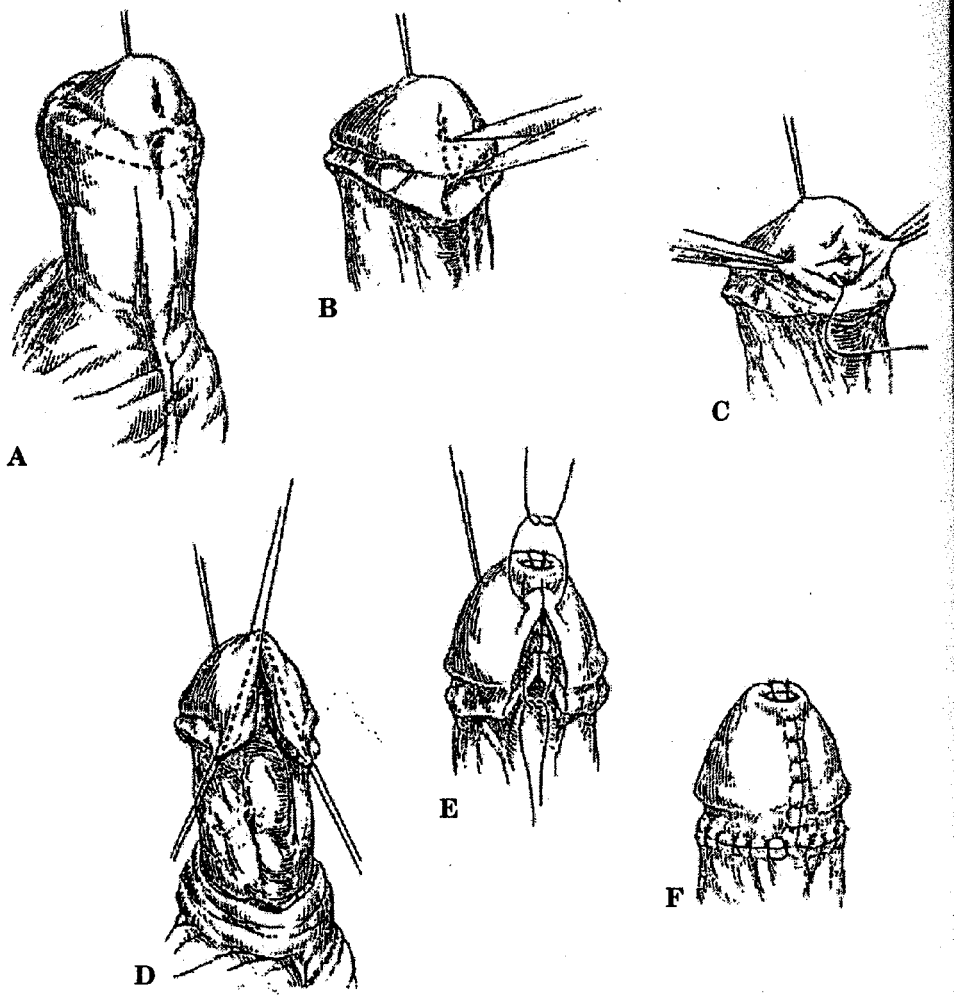


Fig. 2–3. A–F: MAGPI hypospadias repair.

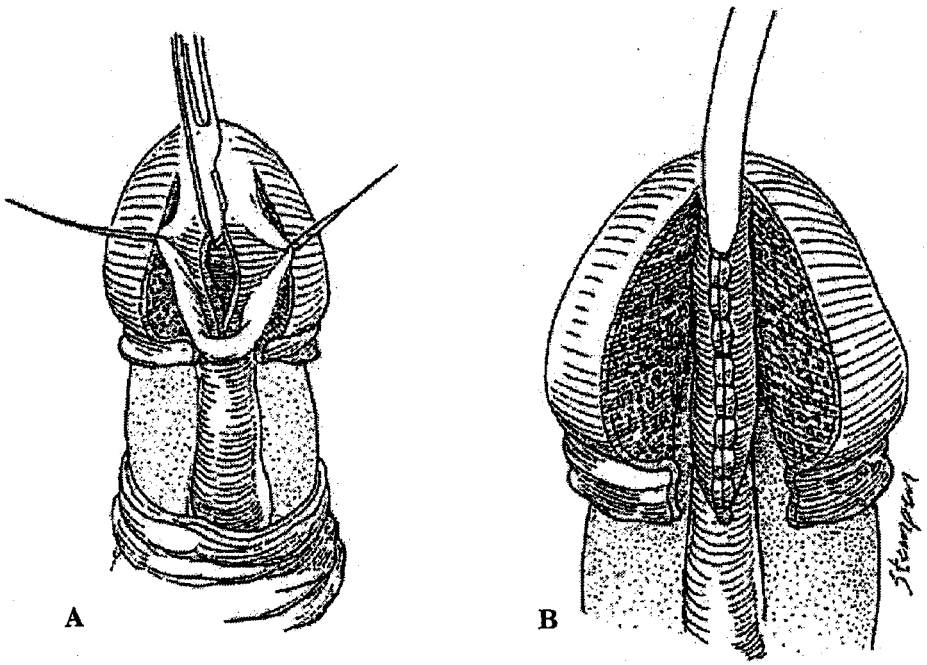
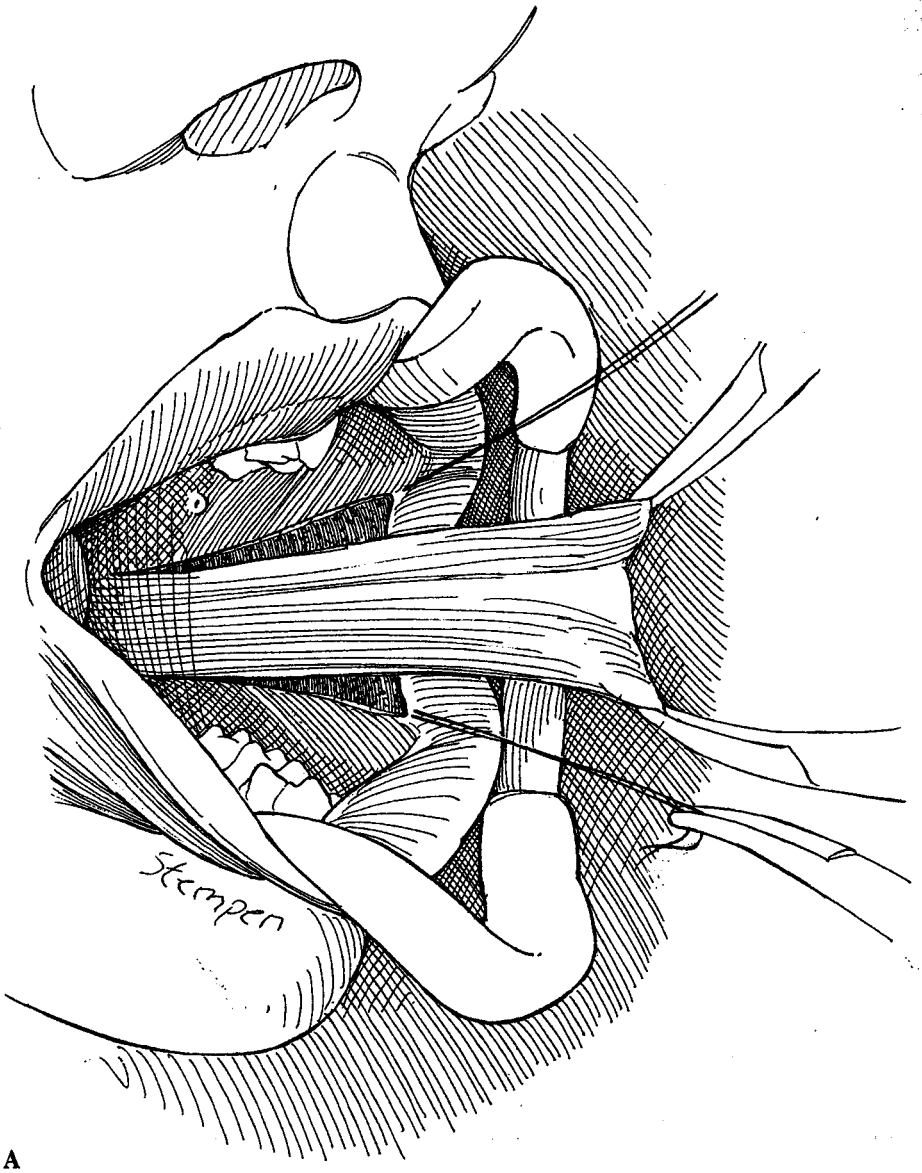
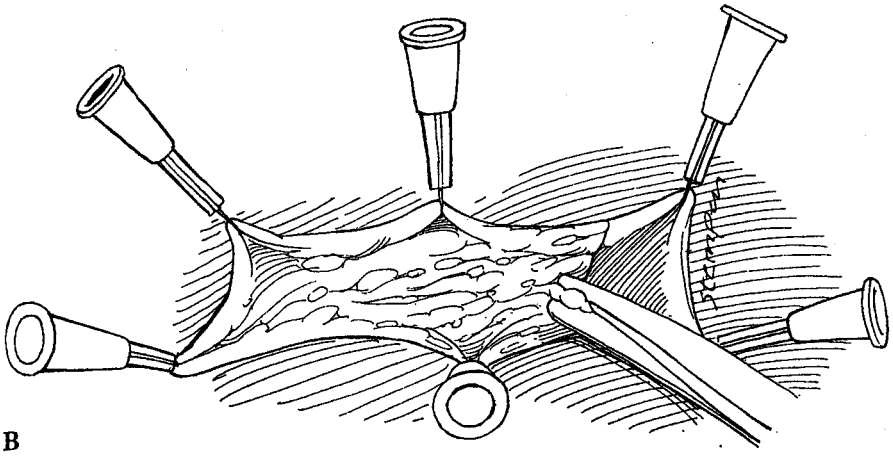


Fig. 2–4. A–B: Snodgrass hypospadias repair.



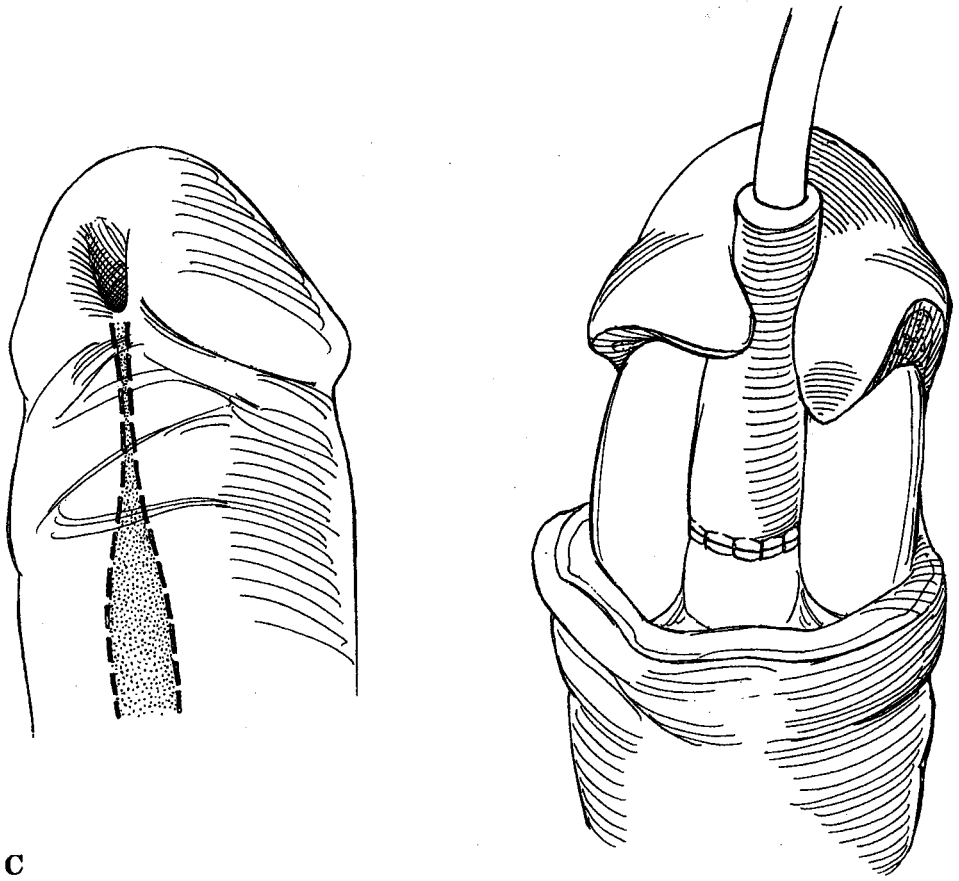
A



B

Fig. 2–6. Buccal mucosa free graft harvest (A), preparation (B), and tubularization

Fig. 2–7. Dorsal plicati



C

Fig. 2–6. (*Continued*) **(C) into new urethra for secondary hypospadias procedures.**

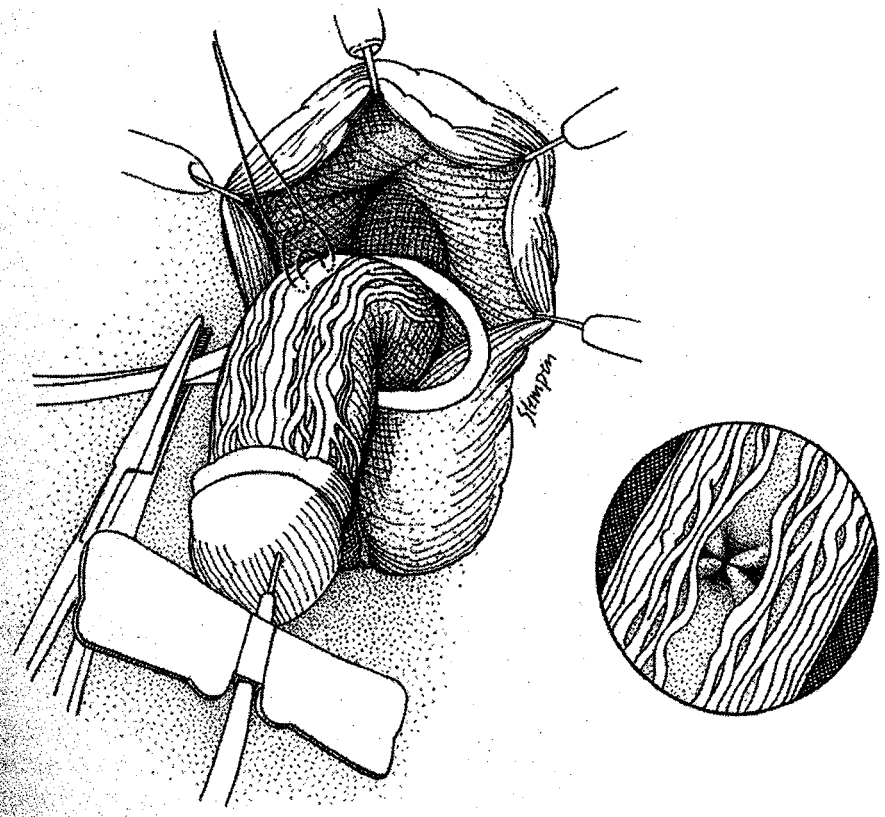


Fig. 2–7. Dorsal plication for the correction of penile curvature.

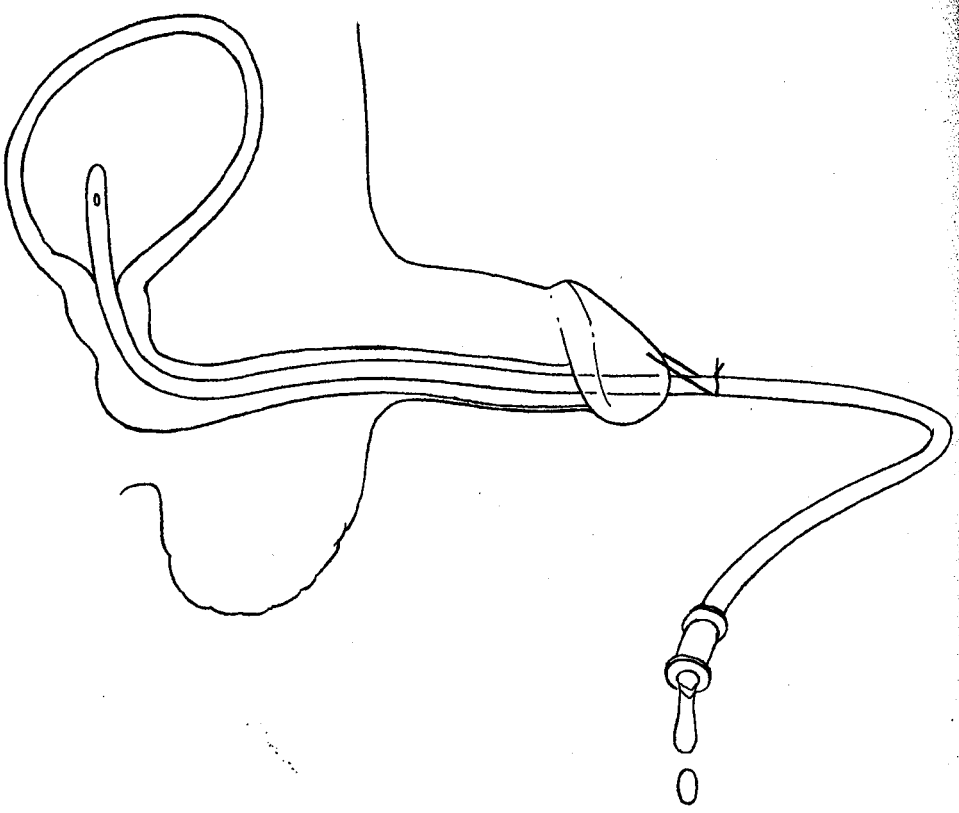


Fig. 2–8. Urinary diversion drippy stent for hypospadias repair.

spontaneously without the need for further surgical intervention by secondary penile skin granulation. This can be treated with local wound care and daily bathing.

E. Residual Penile Curvature: If severe, reoperation with penile straightening is required.

XI. POSTOPERATIVE CARE

(See pediatric patient handout Web site Chapter 31.)

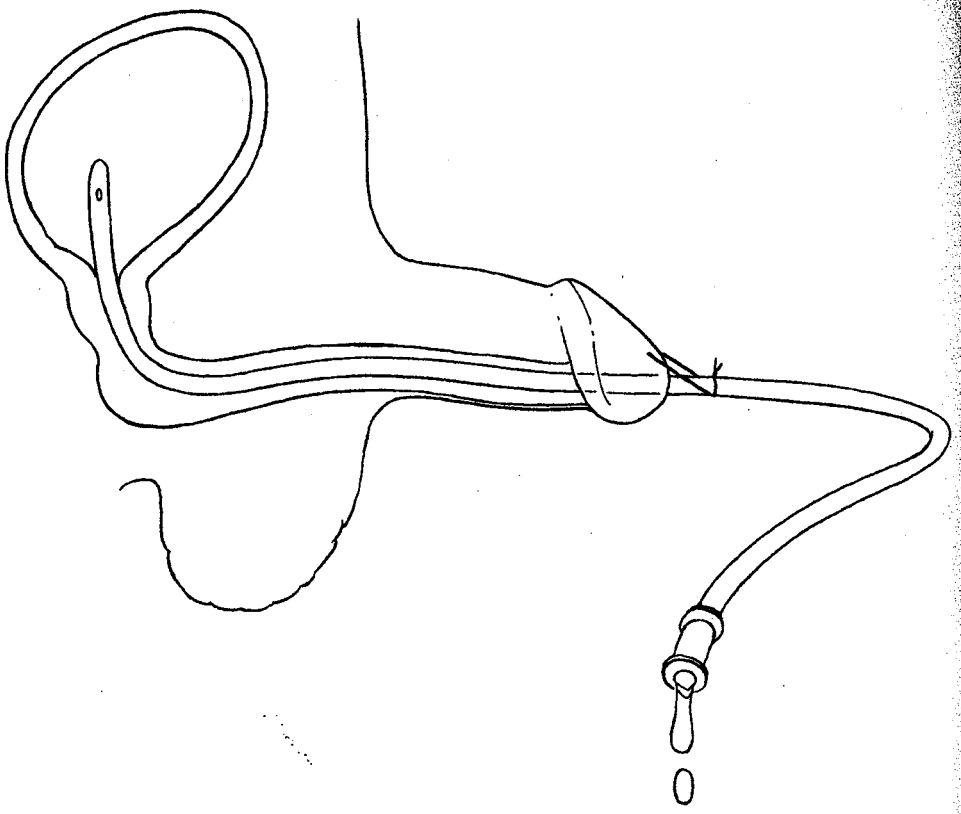
A. Anterior hypospadias repair such as the MAGPI and GAP are often done without the use of a drippy stent and require no special treatment.

B. More severe hypospadias requires the use of an indwelling drippy stent, which is typically removed 7 to 14 days after surgery by cutting a stitch that secures the urethral catheter to the glans penis (Fig. 2–8).

C. Prophylactic doses of antibiotics (see Chapter 32) such as cotrimoxazole (Bactrim) or nitrofurantoin are typically prescribed while the stent is in place to keep the urine sterile.

D. Postoperative symptoms include

1. Bladder spasms, which can be treated with oxybutynin (Ditropan)
2. Urinary retention (uncommon); secondary to a stent malfunction such as blockage or kinking
3. Postoperative pain, controlled with acetaminophen (Tylenol), acetaminophen with codeine elixir, and antiinflammatory agents such as ibuprofen (Motrin)



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E. Dressings
The most common dressing used after hypospadias is a plastic dressing such as Tegaderm, which is used with gauze to sandwich the penis onto the abdomen and is typically removed at home 2 to 3 days after surgery.

XII. POSTOPERATIVE CARE

A. The majority of patients with hypospadias can expect excellent outcomes with one operation. The reconstruction tends to grow with the child and results in a normal phallus for voiding, appearance, and sexual function after puberty.

B. To ensure patient satisfaction and an understanding of their congenital anomaly, my practice is to see patients who have had hypospadias reconstruction 1 year after the repair, after toilet teaching, and after puberty.

C. Patients with complications are seen as needed. With better primary surgical techniques, the goal is to eliminate the need for salvage surgery.

RECOMMENDED READING

Baskin LS. Hypospadias. In: *Pediatric surgery,* 4th ed. O'Neill, 2002.
Baskin LS. Hypospadias. In: O'Neill JA Jr, Rowe MI, Grosfield JL, et al, eds. *Pediatric surgery,* 5th ed. New York: Elsevier, 2004.

7

Vesicoureteral Reflux

Laurence S. Baskin

I. INTRODUCTION

Vesicoureteral reflux is the abnormal retrograde flow of bladder urine into the upper urinary tract through an incompetent ureterovesical junction. Reflux in itself that is without bacterial contamination and low in pressure has not been documented to be deleterious. Reflux in the presence of bacteria is a risk factor for upper urinary tract infections or pyelonephritis. Untreated upper urinary tract infections have been shown to lead to acquired renal scarring or reflux nephropathy in children. Congenital abnormalities of renal development may have associated vesicoureteral reflux, resulting in a clinical picture of reflux nephropathy but without a history of urinary tract infection.

A. Reflux Nephropathy is defined as the following clinical triad.

1. Renal scarring
2. Hypertension
3. Vesicoureteral reflux

II. EPIDEMIOLOGY

The prevalence of reflux varies with several demographic factors of the patient population. Reflux may occur as an isolated entity or with other associated anomalies of the genitourinary tract.

A. History of Urinary Tract Infection

In children without urologic symptoms or history of infection, the incidence of reflux is likely less than 1%. In children with a history of symptomatic urinary tract infection, the incidence of reflux has been estimated to range from 20% to 50%.

B. Age

The prevalence of reflux correlates inversely with the age of the study population. With linear growth, spontaneous resolution of reflux occurs in many patients.

C. Race

Reflux is more commonly a disease of fair-skinned children. The prevalence of reflux appears to be significantly lower among black children and children of Mediterranean origin when compared to white children. The prevalence of reflux in black children with a history of urinary tract infection is estimated to be approximately 25% of the incidence in the population of white children.

D. Sibling Predisposition

Siblings of patients with known reflux have approximately a 30% prevalence of reflux, with younger siblings being at greatest risk. In many of these children,

there may be no documented history of symptomatic infections. For this reason, routine screening of the siblings of children with reflux grade 3 or higher has been advocated by most pediatric urologists and nephrologists.

E. Gender

Because of the epidemiology of urinary tract infection in children, boys and girls may present with reflux at different ages. Because urinary tract infections are more common in uncircumcised boys than girls during the neonatal period, many boys are diagnosed with reflux in the neonatal period. However, after the first year of life, the incidence of urinary infections is much higher among girls than boys. Therefore, most school-aged children diagnosed with reflux are girls.

F. Associated Anomalies

Although vesicoureteral reflux may occur as an isolated entity, reflux may also occur with other genitourinary abnormalities.

1. Posterior urethral valves: Congenital bladder outlet obstruction has been associated with reflux in up to 50% of patients.
2. Duplicated collecting system: Reflux is commonly associated with the lower pole moiety of a duplicated system.
3. Prune-belly (Eagle-Barrett) syndrome
4. Bladder exstrophy
5. Severe voiding dysfunction

III. CLASSIFICATION OF REFLUX

A. Primary Reflux

Primary reflux occurs as a result of a congenital deficiency in the formation of the ureterovesical junction in the absence of any other predisposing pathology. Accordingly, these patients may have a laterally ectopic ureteral orifice consistent with a deficient submucosal ureteral tunnel. The majority of otherwise healthy children who present with symptomatic urinary tract infection have primary reflux. It should be noted that the majority of the current guidelines which have been developed to treat children with reflux, apply mainly to those patients with primary reflux.

B. Secondary Reflux

Secondary reflux occurs as a result of other urinary tract dysfunction, which leads to a decompensation of a normally formed ureterovesical junction. Although secondary reflux may have many different underlying etiologies, the key to the treatment of these patients depends on the identification of these underlying etiologies. Successful treatment of these patients ultimately depends on the management of the underlying causes.

1. Neurogenic bladder
   a. Myelomeningocele
   b. Spinal cord injury

Table 7–1. Grading of vesicoureteral reflux

Grade 1: Appearance of contrast in the ureter only
Grade 2: Appearance of contrast in the ureter and renal pelvis without associated dilation or blunting of calyces
Grade 3: Mild calyceal dilation without ureteral tortuosity
Grade 4: Moderate calyceal dilation and blunting without ureteral tortuosity
Grade 5: Severe calyceal dilation with ureteral tortuosity

2. Obstruction
   a. Voiding dysfunction
   b. Posterior urethral valves
   c. Ectopic ureteroceles
3. Infection: Cystitis may also predispose an otherwise marginally competent ureterovesical junction to demonstrate reflux.

IV. GRADING OF REFLUX

Grading of the degree of reflux is important to the management and ultimate prognosis of the patients. More severe grades of reflux are associated with lower rates of spontaneous resolution and a higher incidence of renal scarring. According to the international system, reflux is graded by the severity of calyceal changes, ureteral dilation, and redundancy that occur secondary to reflux (Table 7–1, Fig. 7–1).

V. CLINICAL PRESENTATION

A. Urinary Tract Infection

Most children with reflux initially present with an episode of urinary tract infection. Pyelonephritis is the initial presentation in most cases.

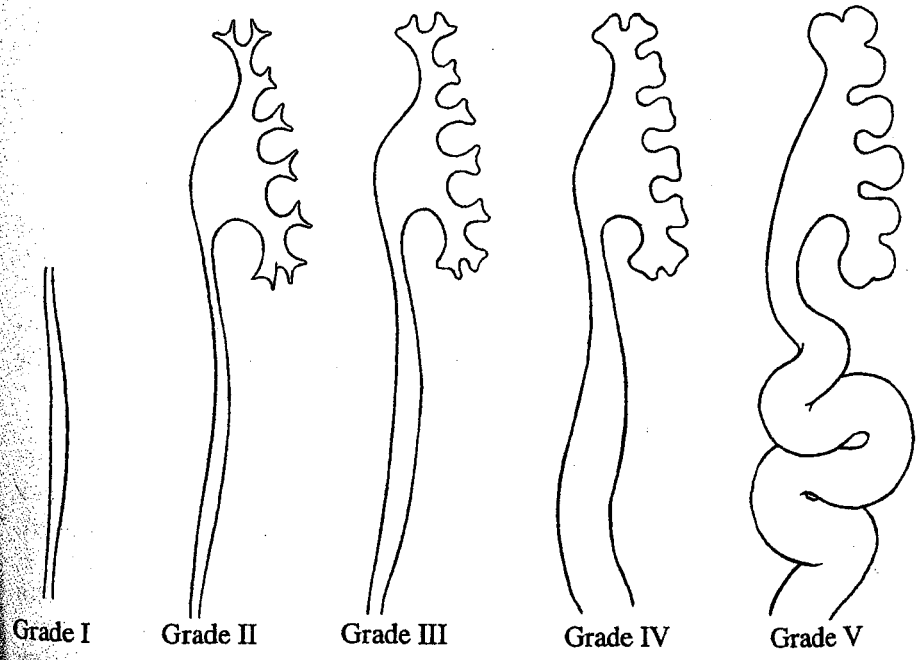

Fig. 7–1. International reflux classification.

B. Unexplained Febrile Illness

Other patients may have no documented history of urinary tract infection, but may instead have a history of frequent recurrent illness with unexplained fevers. Some children with reflux and pyelonephritis are mistakenly treated for presumed recurrent otitis. It is, therefore, of utmost importance to evaluate all children with unexplained fever with a urinalysis and urine culture.

C. Associated Genitourinary Anomalies

As mentioned, reflux commonly occurs in conjunction with other urologic abnormalities. The finding of any of the previously mentioned conditions should prompt an investigation for reflux.

D. Prenatal Diagnosis

Because of the recent advent and popularity of prenatal ultrasound screening, fetal hydronephrosis can often be detected in utero. Although fetal hydronephrosis can be due to any number of pathologic or physiologic causes (see Chapter 12), all children with prenatally diagnosed hydronephrosis should be placed on antibiotic prophylaxis until a cystographic examination can be done to either confirm or exclude reflux.

VI. DIAGNOSIS

Once the acute illness is resolved, the diagnosis of reflux can be entertained. A voiding cystourethrogram (VCUG) should never be performed in the presence of infected urine. However, the likelihood of detecting reflux is highest if the child is evaluated soon after the acute infection episode. It is believed that the presence of recent infection in the bladder predisposes a marginally competent ureterovesical junction to reflux. The presence of reflux in these marginally competent ureterovesical junctions may be missed if the evaluation is conducted several weeks following the acute episode. Because the real danger is vesicoureteral reflux in the presence of infection, many authorities have advocated early radiographic evaluation at 1 to 2 weeks following the acute episode.

A. VCUG

Vesicoureteral reflux is a radiographic diagnosis. The gold standard study is contrast VCUG. VCUG allows accurate diagnosis and grading of the severity of reflux. In a small number of cases, more than one cystographic examination may be needed to make the diagnosis of reflux.

B. Urodynamic Evaluation

Many children with reflux also have voiding dysfunction. Often the diagnosis is suspected by a history of incontinence, frequency, or urgency. If indicated, urodynamic evaluation with monitoring of intravesical compliance can be done at the same time as the VCUG. This information is important in the management of patients with reflux and other abnormalities of the lower urinary tract (e.g., neurogenic bladder).

Appropriate management of the voiding dysfunction often results in resolution of reflux.

C. Renal Ultrasound

Renal ultrasound is relatively insensitive in the detection of mild and moderate reflux. As an adjunctive technique, ultrasound can be useful in the grading of hydronephrosis (see Chapters 12 and 13), and as a baseline for follow-up studies to monitor renal growth.

D. Nuclear Medicine Renal Scan

Renal scan using DMSA is useful in the detection of renal cortical scars. Some centers have employed renal scans to confirm the diagnosis of acute pyelonephritis.

E. Nuclear Medicine Cystogram

Nuclear cystogram is useful as a follow-up study in patients with known reflux. It has the advantage of high sensitivity and lower radiation exposure than the standard VCUG. However, because of its inability to accurately grade reflux and detect associated anomalies (spina bifida, ureteroceles, duplicated systems), nuclear cystograms should not be used for the initial evaluation of reflux.

VII. MANAGEMENT

The primary goal in both medical and surgical management of reflux is to prevent development of pyelonephritis, recurrent urinary tract infections, and the formation of renal cortical scarring.

A. Medical Management of Reflux

Once the diagnosis of reflux is suspected, low-dose continuous antibiotic prophylaxis should be initiated. The patient should continue antibiotic prophylaxis until the reflux resolves spontaneously or is corrected surgically. Each patient should have periodic follow-up (yearly to 18 months) to check for the resolution of reflux.

1. Antibiotic prophylaxis
   There are relatively few effective antibiotic agents available for use in urinary tract prophylaxis in children. Prophylactic antibiotics are generally given in a dosage that is approximately one-half to one-third of the normal therapeutic dose. Table 7–2 lists the commonly used agents and potential serious-but-rare side effects.
2. Anticholinergic therapy
   For patients with uninhibited bladder contractions (dysfunctional voiding; see Chapter 8) and secondary reflux, treatment with anticholinergic medications (e.g., oxybutynin) in addition to antibiotics may allow spontaneous resolution to occur.
3. Office visits and follow-up testing
   Patients on medical management should be under surveillance for urinary tract infections. However, routine monitoring of urine is not

Table 7–2. Commonly used agents for urinary tract prophylaxis in children

Agent	Oral Dosage	Precautions
Trimethoprim/ sulfamethoxazole	2 mg/kg PO qhs (Trimethoprim)	Avoid use in the first 2 months due to kernicterus, megaloblastic anemia.
Nitrofurantoin	2 mg/kg qd	Pulmonary fibrosis, hemolytic anemia in G6PD deficiency.
Amoxicillin	20 mg/kg PO qd	Good choice for first 2 months. Development of resistance common thereafter.
Cephalexin	20 mg/kg PO qd	Good choice for first 2 months. Development of resistance common thereafter.

recommended unless the patient has signs and symptoms of a urinary tract infection. Urine should periodically be monitored for protein and the patient checked for hypertension, although in most cases proteinuria and hypertension will not be an issue until well into adulthood. Kidney growth as well as scarring should be monitored with ultrasound (every 1 to 2 years). The critical issue is to keep the urine sterile while reflux is present, thereby decreasing the chance of pyelonephritis and new renal scarring.

B. Surgical Correction

Although many patients with reflux can be managed successfully with antibiotic prophylaxis, surgical correction of reflux is often indicated when medical therapy is unsuccessful.

1. Indications
   - **a.** Recurrent episodes of pyelonephritis on antibiotic prophylaxis
   - **b.** Medical noncompliance
   - **c.** Breakthrough infections by resistant organisms
   - **d.** Persistence of reflux into puberty
2. Results

   In experienced hands, antireflux surgery is highly safe and effective. In the American section of the International Reflux Study Group, ureteral reimplantation successfully corrected reflux in 99% of cases with complication rates less than 2%.

3. Techniques
   a. Open surgery: The goal of surgery is to create a functional value at the vesicoureteral junction. This is accomplished by mobilization of the ureter and reimplantation using stronger muscular backing. Two general approaches are used intravesical (Fig. 7–2) and extravesical (Fig. 7–3). As noted, surgical repair has an extremely high success rate.

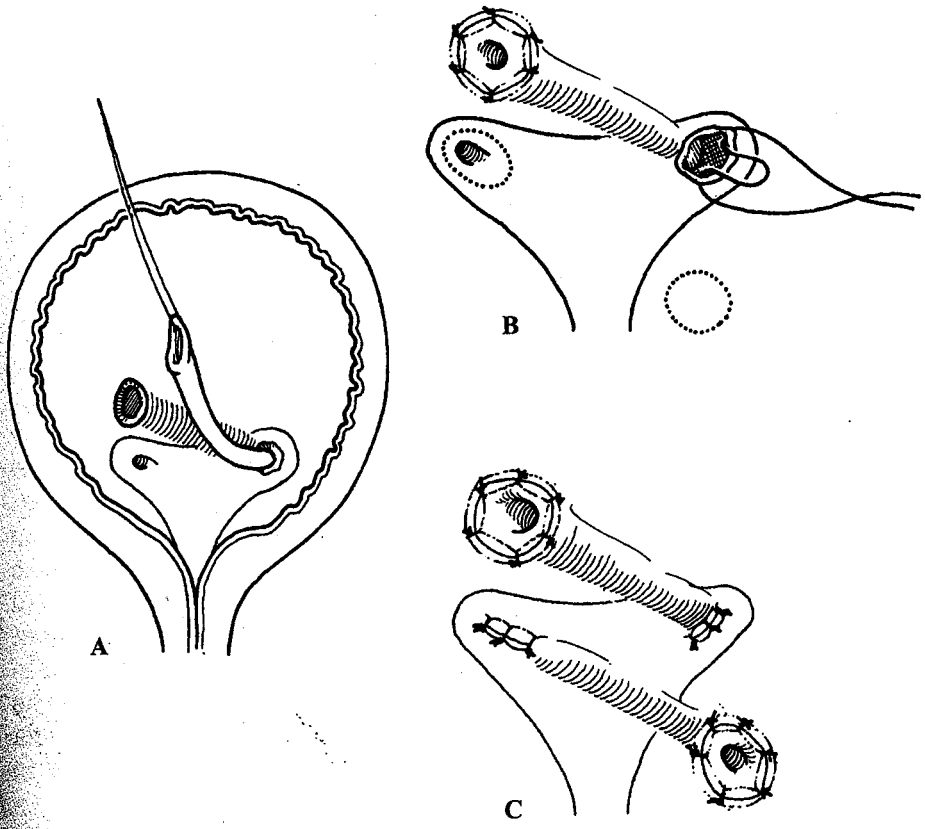

Fig. 7–2. A–C: Intravesical ureteral reimplantation technique.

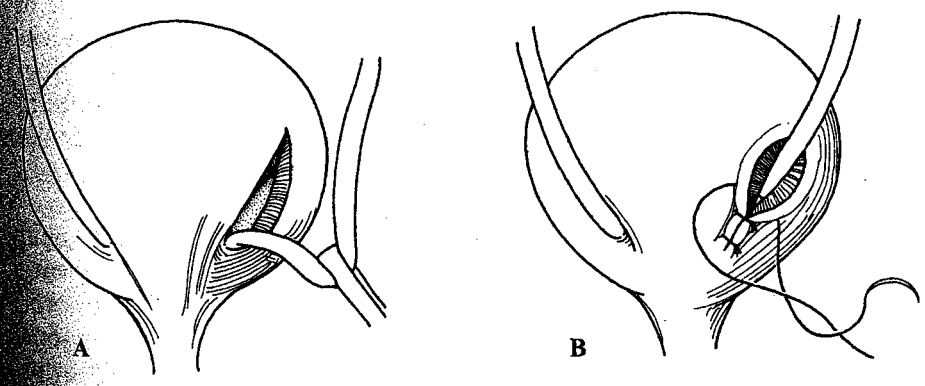

Fig. 7–3. A–B: Extravesical ureteral reimplantation technique. Posterior aspect of the bladder (Lich-Gregor procedure).

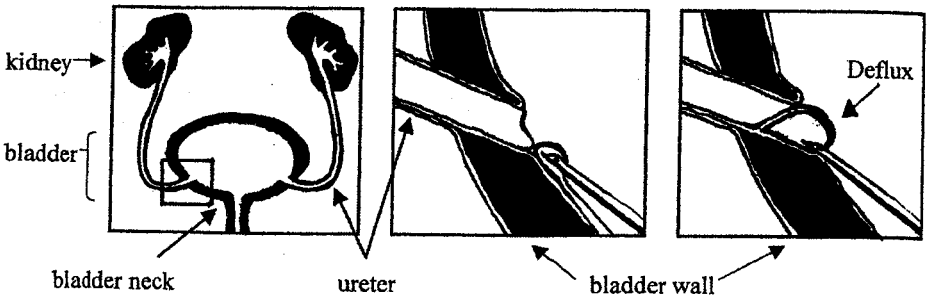

Fig. 7–4. Endoscopic injection of dextranomer/hyaluronic acid copolymer for the correction of vesicoureteral reflux.

b. Endoscopic treatment

(1) Injectable agents

The endoscopic treatment of vesicoureteral reflux has become accepted in the United States with the FDA approval in 2001 of injectable dextranomer/hyaluronic acid copolymer (Deflux). The procedure is performed by inserting a cystoscope, through the urethra to access the inside the bladder (Fig. 7–4). A small amount of the injectable copolymer is introduced into the wall of the bladder near the opening of one or both ureter(s). Although the exact mechanism of reflux correction is unknown, the copoloymer seems to fix the ureterovesical junction in place and recreate the natural intravesical tunnel between the ureter and bladder. There are no incisions made in the abdomen for this procedure. Including the administration of general anesthesia in children, the procedure takes 15 to 30 minutes.

(2) Risks

The use of this injectable substance has minimal risks. These include mild bleeding, infection, and dysuria after the procedure. The theoretical risk of ureteral obstruction and kidney blockage has been reported only rarely. Nevertheless, follow-up upper tract imaging with an ultrasound 4 to 6 weeks after the procedure is prudent.

(3) Outcomes

Using the dextranomer/hyaluronic acid copolymer, there is a higher success rate with this procedure for those with lower grades of reflux. Capozza and Caione (2002) reported a 95% success rate for grade II, 71% success rate for grade III,

and 43% success rate for grade IV reflux. Similarly, in their study Capozza et al. (2001) reported an 87% success rate for grade II, 75% success rate for grade III, and 41% success rate for grade IV reflux. In another study, Lackgren et al. (2001) reported a 78% success rate for both grades II and III reflux, and 66% success rate for grade IV reflux. The treatment can be performed more than once and does not impede open surgical correction if not successful.

(4) Candidates for This Procedure
Based on the success rates, this procedure is recommended for use in children with grade II, grade III, and possibly grade IV reflux.

(5) Contraindications
- **(a)** A large bladder diverticulum
- **(b)** An extra ureter (tube that carries urine from the kidney to the bladder)
- **(c)** Active urinary tract infection
- **(d)** Active voiding dysfunction (abnormal emptying of the bladder)

VIII. SUMMARY

Vesicoureteral reflux is one of the most common urologic abnormalities treated by pediatric urologists. An increased understanding of the pathologic mechanisms, bladder dysfunction, and natural history of reflux has led to improvements in both medical and surgical approaches to treating this disease. The effective treatment of patients with reflux depends on early detection of reflux and institution of antibiotic prophylaxis. Close follow-up and periodic reevaluation are necessary for all patients. When indicated, surgical correction of reflux has been shown to be safe and effective. Endoscopic treatment with agents such as the dextranomer/hyaluronic acid copolymer has proven to be safe and effective alternative treatment.

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12

Prenatal Urologic Diagnosis and Maternal Counseling

Barry A. Kogan

With the advent of the widespread use of prenatal ultrasound in clinical practice, urologic conditions are being diagnosed in many infants before birth. Because ultrasound is performed so commonly, the impact of these diagnoses has been enormous. It remains unclear, however, whether this early diagnosis results in benefit to the neonate or merely increased anxiety over testing. In the majority of cases, parents can be reassured that postnatal evaluation is all that is needed and that their infant will have a good prognosis.

I. INCIDENCE

Urologic anomalies are found in about 0.4% of pregnancies. Of these, hydronephrosis is found in about half (see Chapter 13).

II. FINDINGS

Urologic findings may range from mild hydronephrosis that is essentially a normal finding to significant hydronephrosis resulting from any type of obstructive uropathy (Figs. 13-1, 13-2, and 13-3). In rare cases, renal tumors can be found or congenital anomalies like exstrophy and epispadias.

III. GENERAL TREATMENT

After initial diagnosis, a more extensive ultrasound survey is performed to confirm the diagnosis, look for associated findings, and, in the case of renal anomalies, to monitor the amount of amniotic fluid. In a few cases, some intervention is warranted in utero; for example, aspiration of an enormous cystic mass or, in very rare cases, placement of a vesicoamniotic shunt. It is almost never indicated to deliver the baby early. In most cases no intervention is needed before birth. In the majority, a postnatal evaluation is recommended, as well as prevention of urinary tract infection prior to postnatal evaluation. However, the overall benefits of prenatal diagnosis are unproven.

IV. SPECIFIC DIAGNOSES

A. Hydronephrosis: Easy diagnosis in utero. The following parameters are important.

1. The amount of amniotic fluid (correlates loosely with renal function)
2. Unilateral or bilateral (unilateral less concerning because of the normal contralateral kidney)
3. Evaluate the amount of hydronephrosis [generally by measuring the anteroposterior (AP) diameter of the renal pelvis]. An AP diameter greater than

10 mm of the renal pelvis in the third trimester warrants a postnatal evaluation (see below).

4. Amount of renal parenchyma
5. Corticomedullary differentiation (when present, suggests good renal function)
6. Echogenicity (when present this suggests renal dysplasia)
7. Cortical cysts (when present this suggests renal dysplasia)
8. Ureteral dilation (suggests a problem at the bladder or urethral level) and requires a postnatal evaluation
9. Bladder distention (suggests a problem at urethral level)
10. These diagnoses rarely, if ever, require in utero treatment. Another example of the very rare scenario where intervention would be considered is a massively obstructed solitary kidney from an ureteropelvic junction obstruction with progressive decrease in amniotic fluid.
11. Postnatally, patients with significant prenatal hydronephrosis as defined by an AP diameter greater than 10 mm in the third trimester or ureteral dilation should have a sonogram and a voiding cystourethrogram (VCUG; 33% incidence of reflux) at several weeks of age.
12. Prophylactic antibiotics [amoxicillin or cephalexin (Keflex)] should be given until proven that there is no reflux.
13. As a corollary, a prenatal diagnosis of hydronephrosis with an AP diameter less than 10 mm in the third trimester rarely if ever results in a clinically significant postnatal outcome such as loss of renal function, pain, or progressive hydronephrosis. These patients have a slightly dilated yet normal functioning kidney. They do not need postnatal evaluation unless clinical suspicion such as a urinary tract infection, hematuria, or pain results.
14. Circumcision is not contraindicated for boys with significant prenatal hydronephrosis. It may be beneficial because it may reduce the rate of urinary tract infections.
15. Generally, patients with prenatal hydronephrosis have an excellent prognosis.

B. Urethral Valves

1. Diagnosed by hydronephrosis, bladder distention, and the key-hole sign on ultrasound. The same findings as above are important, but especially amniotic fluid and renal parenchymal changes.
2. If these are okay, then the most appropriate treatment is to defer treatment until after birth.
3. At that time the neonate should have catheter drainage, VCUG, and resection of valves if he is

large enough; alternatively, a temporary vesicostomy can be used to bypass the blockage.

4. In rare cases, consider fetal intervention (see below).
5. Prognosis depends on the degree of renal dysplasia and is generally determined after birth if amniotic fluid is normal.

C. Multicystic Kidney

1. Easily diagnosed in utero. The main findings are chaotic configuration of the kidney, multiple cysts that do not communicate, and no normal renal parenchyma.
2. Generally associated with a normal contralateral kidney and normal amniotic fluid.
3. Postnatally, we recommend prophylactic antibiotics until ultrasound and VCUG (25% of children have contralateral reflux into the solitary kidney). This condition will generally not affect life expectancy or health. In most institutions, nephrectomy has become rare as most of these kidneys involute on their own.

D. Duplications, Ectopic Ureters, or Ureteroceles

1. Often diagnosed in utero by hydronephrosis of one pole of a kidney (typically the upper pole), ureteral dilation, and/or a classic ureterocele configuration in the bladder.
2. Provided amniotic fluid is normal, no further in utero treatment is needed. Prophylactic antibiotics and postnatal ultrasound and VCUG are appropriate (see Chapter 14 for treatment).

V. PRENATAL TREATMENT AND MATERNAL COUNSELING

A. Urinary Tract Obstruction

1. Prenatal intervention is appropriate in only the exceptional urologic case.
2. The most common circumstance is in cases of severe bilateral hydronephrosis with reduced amniotic fluid. In those instances, a careful fetal survey (to rule out other anomalies) and amniocentesis (for chromosome analysis) are essential before consideration of intervention.
3. Analysis of the degree of renal injury is the next step and this is done first by ultrasound. In general, acceptable renal function is associated with normal amniotic fluid; however, when the fluid is reduced other assessments are needed.
4. The amount of renal parenchyma can be assessed easily. More specifically, though, the lack of corticomedullary differentiation and the presence of increased echogenicity and cortical cysts are all associated with renal dysplasia.
5. If the kidneys are acceptable by ultrasound, fetal urine samples are obtained (some feel it is best to drain the bladder 3 days in a row to obtain fresh urine).

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6. Although many tests have been proposed, low sodium and low osmolarity (indicative of enough tubular function to reabsorb urine) are associated with salvageable renal function.
7. In these highly selected patients, after counseling of maternal as well as fetal risks, consideration may be given to in utero intervention.
8. Although there are reports of fetal cystoscopic treatment, the accepted intervention is a fetal vesicoamniotic cavity shunt (usually via percutaneous catheter placement). This is done under local anesthesia with maternal sedation and with careful monitoring and medications to reduce the risk of premature delivery. Despite this, many of these infants do deliver early.
9. Long-term results are mixed. There is a clear benefit in preventing pulmonary hypoplasia (and thereby increasing neonatal survival), but it is still unclear whether renal functional improvement is achieved. This brings up the ethical concern that these children may be surviving due to therapy, but doomed to neonatal renal failure with all its attendant problems.

B. Myelodysplasia

1. Prenatal diagnosis of this condition is increasing owing to increased use of maternal screening for α-fetoprotein.
2. Based on this, prenatal open surgical hysterotomy and fetal myelomeningocele repair is being done in several centers as part of a multiinstitutional study sponsored by the National Institutes of Health.
3. Early results suggest that the rate of ventriculoperitoneal shunting is reduced in these patients, but neurogenic bladder dysfunction is still a problem. However, these results are still too early and too limited to draw firm conclusions.

I. SUMMARY

A. Fetal diagnosis of urologic anomalies is common.

B. Although the benefits are not proven fully, the prevalence of prenatal ultrasound means that many children will be diagnosed.

C. In most instances, parental education should emphasize the excellent prognosis and the need for nonemergent postnatal evaluation.

D. In extremely rare cases, fetal intervention may be warranted.

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13

Neonatal Hydronephrosis (UPJ Obstruction) and Multicystic Dysplastic Kidneys

Ahmet R. Aslan

NEONATAL HYDRONEPHROSIS (UPJ OBSTRUCTION)

I. DIAGNOSIS

The diagnosis of neonatal hydronephrosis is most often made based on an incidental finding detected on prenatal ultrasound done for another reason. Occasionally, an infant may be brought to the physician with a flank mass, urinary tract infection, or rarely with hematuria. Older children most often present with intermittent pain and vomiting.

A. Incidence

1. The incidence of hydronephrosis varies depending on the cut-off level for renal anteropelvic diameter that is used during prenatal sonographic evaluation.
2. Because of high false-positive rates, pelvic diameters below 10 mm are generally not considered significant.
3. Although the incidence of some renal enlargement on prenatal ultrasound is 1.0% to 1.4%, most of these cases resolve after birth and the incidence of significant urinary tract disease is 0.2% to 0.4%.

B. Differential Diagnosis of Prenatal Urinary Tract Dilation (Fig. 13–1)

1. Ureteropelvic junction (UPJ) tract hydronephrosis (UPJ Obstruction) (about two-thirds of cases)
2. Vesicoureteral reflux (about one-fourth of cases)
3. Ureterovesical junction (UVJ) obstruction (will have a dilated ureter)
4. Multicystic dysplastic kidneys (multiple renal cysts that do not connect to each other)
5. Posterior urethral valves (will have a markedly abnormal bladder also)
6. Other diseases like prune-belly syndrome or polycystic kidney disease (rare)

C. Etiology of UPJ Obstruction (Fig. 13–2)

1. The exact cause is debatable, but a scarred or adynamic segment of the proximal ureter often leads to a kink at the UPJ that may be responsible for the obstruction.
2. Less common causes are congenital mucosal folds or polyps of the upper ureter or in older children a crossing artery to the lower pole of the kidney. Figure 13–2A compares UPJ type hydronephrosis to ureterovesical junction (UVJ) type hydronephrosis 13–2B.

Etiology of Hydronephrosis
Multi Cystic Dysplastic Kidney
Posterior Urethral Valves
Dysplastic Kidney
Normal Post-Natal Imaging
Vesico-Ureteral Reflux
UVJ Obstruction 13.4%
UPJ Obstruction 64.0%

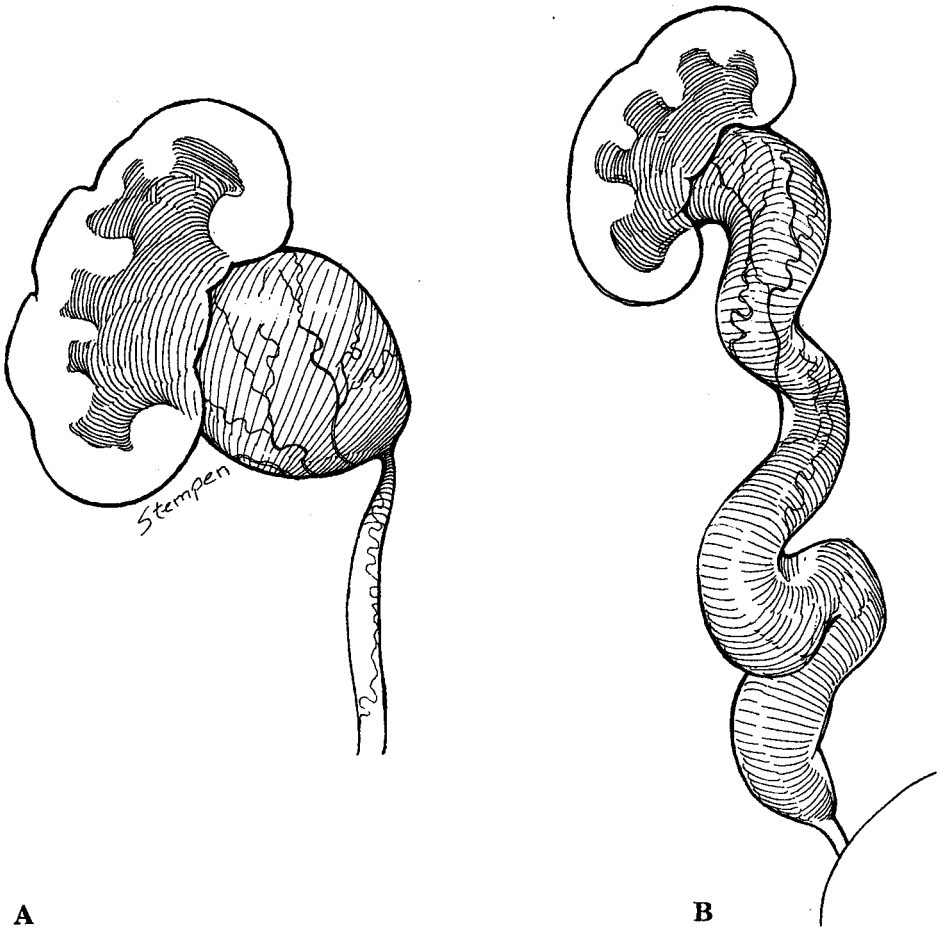

Fig. 13–2. Schematic of ureteropelvic junction obstruction (A) as compared to congenital megaureter (B).

Fig. 13–1. Common etiologies of hydronephrosis in children.

II. EVALUATION OF NEONATAL HYDRONEPHROSIS

A. Ultrasound

1. Unless there is severe bilateral hydronephrosis, dilated ureter, or dilated posterior urethra on prenatal sonograms, postnatal ultrasound should be postponed until the physiologic dehydration of newborn has resolved (usually after 72 hours). Earlier testing may underestimate the degree of hydration.
2. The grading system of the Society of Fetal Urology may be used as a guideline to determine the degree of hydronephrosis (Fig. 13–3 and Table 13–1). Another way to define hydronephrosis is to simply measure the greatest dimension of the renal pelvis. As mentioned, renal pelvic diameters greater than 10 mm (grade 2 on the Society of Fetal Urology grading system) should be considered clinically relevant and require further evaluation. Clearly, quantification of the hydronephrosis is dependent on the ultrasound operator and the hydration status of the patient. The status of the urinary bladder can also affect the degree of hydronephrosis (when

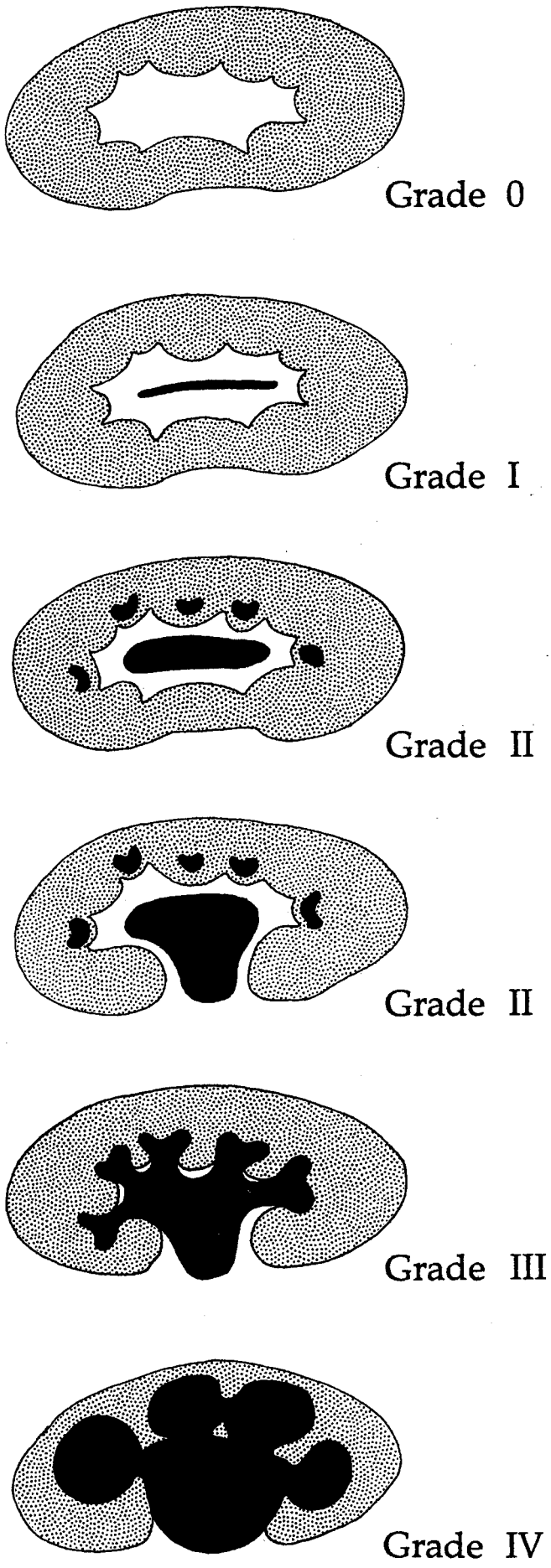

Fig. 13-3. Ultrasound grading system of hydronephrosis (see text).

Table 13–1. Ultrasound grading system for hydronephrosis

Grade of Hydronephrosis	Central Renal Complex	Renal Parenchymal Thickness
0	Intact	Normal
1	Slight splitting	Normal
2	Evident splitting Complex confined Within renal border	Normal
3	Wide-splitting pelvis Dilated outside renal Border and calices Uniformly dilated	Normal
4	Further dilation of renal pelvis and calices	Thin

possible, hydronephrosis should be assessed when the bladder is both full and empty.)

B. Voiding Cystourethrography (VCUG)

1. Most clinicians recommend a VCUG in every infant with neonatal hydronephrosis because sonography is not a good predictor of vesicoureteral reflux and 25% to 33% of these children will have reflux.
2. Some will counterargue that this reflux is not clinically significant.
3. We perform a VCUG at 3 to 4 weeks of age and recommend prophylactic antibiotics until the diagnosis of vesicoureteral reflux is ruled out.

C. Radionuclide Scanning

Not all urinary tract dilation is a sign of significant obstruction. Although the size of the renal pelvis is of some value, it is not definitive in most cases. To help distinguish "obstructed" kidneys from those that are merely dilated, the most commonly used test is the diuretic renogram. This test estimates the relative renal function of the two kidneys and gives an estimate of the "washout" of radioisotope from the kidney. This washout correlates with the degree of obstruction (although the size of the renal pelvis and the amount of diuresis are also critical in determining the rapidity of the washout). The test requires hydration and the injection of a radioisotope that is excreted in the urine. The two principle choices are:

1. 99m-technetium-diethylenetriaminepentaacetic acid (DTPA): DTPA is cleared by glomerular filtration and is not reabsorbed or excreted by tubules. Thus, it provides a good measurement of glomerular filtration rate.
2. 99m-Technetium-mercaptoacetyltriglycine (MAG-3): MAG-3 is the first choice in children with poorly

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functioning kidneys because it is mostly cleared by tubular excretion. It is also a good indicator of effective renal plasma flow.

It is important to recognize the technique and related pitfalls. After the administration of radiopharmaceutical agent (DTPA or MAG-3) and taking serial images with a gamma camera, the regions of interest (ROI) over both kidneys and the background are defined. The amount of radioisotope excreted in the urine (the number of counts in each ROI) over time is calculated; then the background activity is subtracted from the kidney counts. Furosemide is administrated 20 minutes after renogram (F+20) to develop time–activity washout curves. The rapidity of washout is correlated with the degree of obstruction. Some authors have tried to use the half-time clearance of radioisotope from the collecting system ($t_{1/2}$) as an indicator of obstruction ($t_{1/2}$ of more than 20 minutes, obstructed; $t_{1/2}$ of 10 to 20 minutes, indeterminate; $t_{1/2}$ of less than 10 minutes, nonobstructed), but there are many problems with the calculation as well as its dependence on the anatomy. $t_{1/2}$ should be interpretted with caution. Overall, the technique is affected by the following items:

a. The degree of obstruction (severe obstruction may cause in delays in peak renal activity with reduction in renal filtration pressure)

b. The compliance of renal pelvis (a highly dilated or compliant pelvis may not respond well to diuretic)

c. The dosage and timing of furosemide (F+20 and F-15 have different effects)

d. The degree of hydration (dehydration may delay excretion)

e. Drainage of the bladder (a full bladder may delay renal washout)

In sum, the data collected in radionuclide scanning must be interpreted depending not only on computer analysis, but also on clinical status of the patient. The test is particularly unreliable before 4–6 weeks of age.

Although not useful in determining the degree of obstruction, DMSA is the most reliable technique to show the relative renal function (it remains bound to proximal tubular cells especially before 1 month of age.)

III. MANAGEMENT OF UPJ HYDRONEPHROSIS AND OBSTRUCTION IN INFANTS

A. Natural History of UPJ Hydronephrosis (conservative management)

The ultimate goal of therapy is to preserve renal function in these children and, at the same time, to prevent unnecessary surgery. Unfortunately, there are no clear-cut methods to predict whether a hydronephrotic kidney will deteriorate or remain unaffected. In fact, the majority of these infants do very well over a long period

of time when observed. However, there are some data to provide guidelines.

1. In children with significant hydronephrosis who are observed, there is a need for future surgery in ~25%.
2. Kidneys with renal pelvic diameter of greater than 50 mm have a high likelihood of renal deterioration; hence, early repair seems to be the best choice for this group.
3. Kidneys with renal pelvic diameter of less than 20 mm rarely need intervention and may be followed using serial ultrasounds and, sometimes, diuretic renography.
4. When the hydronephrotic kidney has a relative renal function less than 40% or has a loss of function greater than 10% in serial scans, surgery is generally recommended.
5. In routine follow-up, there should be a high index of suspicion for urinary tract infections, as there are no specific symptoms in this age group. Infections in the face of obstruction may cause rapid renal deterioration. The use of a prophylactic antibiotic is controversial in cases with isolated hydronephrosis without ureteral dilatation or reflux. We generally do not use antibiotics in these cases.
6. Both infections and stone formation are also indications for surgery.

B. Surgery

1. Anderson-Hynes dismembered pyeloplasty is the gold standard of techniques used for the repair of UPJ with an overall success rate of more than 95%.
2. Endopyelotomy (either balloon cautery or cold knife) is feasible in older children, but the success rate is only about 80% and it is not routinely used in pediatric population. Also it may be particularly dangerous when an extrinsic obstruction (usually an aberrant vessel) is present.
3. Laparoscopic pyeloplasty is becoming a reasonable alternative to the open surgery in pediatric population. In children, however, the advantages of a shorter hospital stay and faster recovery time are small.

MULTICYSTIC DYSPLASTIC KIDNEYS (MCDK)

Multicystic dysplastic kidneys are a severe form of renal dysplasia in which the kidney has virtually no function. On ultrasound, there is very thin and abnormal renal parenchyma, surrounded by multiple cysts of various sizes that do not connect, nor do they connect to the renal pelvis.

I. DIAGNOSIS

Most cases of MCDK are diagnosed incidentally during prenatal sonography. In more unusual cases, it may be

diagnosed after discovery of a palpable or visible abdominal mass in the infants. MCDK must be distinguished from the hydronephrosis caused by UPJ obstruction because the latter may be repairable. The diagnosis may be made by ultrasound with good reliability. The existence of infundibular connections between calyces and the renal pelvis is good evidence of hydronephrosis, instead of MCDK.

II. EVALUATION

A. Renal Scan

There is an increased incidence of UPJ hydronephrosis and obstruction (3% to 12%) in the contralateral (solitary functional) kidney. A radionuclide scan is also recommended if there is any question of UPJ obstruction in the affected kidney. In hydronephrosis, a rim of functioning parenchyma is seen. In MCDK, there is little or no uptake of radionuclide.

B. VCUG

Because there is a high rate of reflux in the contralateral (solitary functioning) kidney (18% to 43%), most clinicians recommend a VCUG.

III. NATURAL HISTORY

A. When multicystic kidneys occur bilaterally, this condition is not compatible with life and these children die immediately after birth from pulmonary hypoplasia.

B. Most cases are unilateral and virtually all become smaller over time. When the involution occurs, the cyst fluid disappears and the dysplastic cells may remain in place; however, in most cases the involution is severe enough that there is generally not any identifiable tissue on ultrasound. It is highly likely that involution occurs in most cases, as the condition is extremely rare in adults.

C. Hypertension occurs in rare cases, so the infant's blood pressure should be checked periodically. When hypertension has been reported, nephrectomy has been curative.

D. The presence of dysplastic cells has lead to the theory that these children have an increased risk of malignancy. In fact the development of Wilms tumor in these patients has been reported only once. Removal of a multicystic dysplastic kidney is rarely indicated with the occasional indication of respiratory compromise from a huge multicystic kidney.

IV. MANAGEMENT

A. When detected prenatally, unilateral multicystic kidney should be followed relatively unaggressively. Unless it is massive in size, there is no need for any change in the routine treatment prenatally or in the mode of delivery.

B. Postnatally, an ultrasound should be obtained to confirm the diagnosis (and a VCUG to be certain there is no contralateral reflux). Once the diagnosis is confirmed, observation is usually recommended. Most clinicians recommend annual sonography; however, there are no objective data to indicate that this is needed. Indeed,

the length of time that a patient with multicystic kidney should be monitored remains unknown. In an era of evidence-based medicine, routine annual ultrasound may be found to be costly and of minimal benefit.

C. Families should be counseled because the child has a solitary functional kidney. It is generally recommended that they avoid contact sports out of concern for injuring the solitary kidney. On the other hand, injuries sufficient to result in nephrectomy are vanishingly rare in contact sports (see Chapter 28).

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16

Congenital Anomalies (Urachal Anomalies, Exstrophy–Epispadias Complex, Imperforate Anus, and Prune-Belly Syndrome)

Ronald S. Sutherland

I. URACHAL ANOMALIES (Fig. 16–1)

A. Background. The *urachus* is a fibrous band that extends from the anterior bladder wall to the umbilicus as the remnant of the allantoic duct. Ordinarily, this tract obliterates by the end of the first trimester; it rarely remains partially or completely open.

B. Presentation. Complete failure of urachal obliteration results in a persistent communication from the bladder with constant leakage of urine. Although this may occur as a result of bladder outlet obstruction such as posterior urethral valves, more often there is no associated anomaly. The usual presentation is a neonate who has a constant wet umbilicus that worsens during crying or straining. If the urachal lumen is only partially obliterated, it may present later in childhood because of enlargement (due to accumulation of desquamated products) or infection. Symptoms and signs include pain, fever, mass, umbilical drainage, and signs of urinary tract infection (voiding frequency, urgency, and dysuria). Included in the differential diagnosis of such a presentation are infected urachal cyst, sinus, or diverticulum; vitelline cyst; umbilical hernia; and ovarian cyst. Usually there is an associated urinary tract infection if there is a communication with the bladder.

C. Workup. If one suspects a urachal anomaly, an ultrasound of the abdomen is usually diagnostic. Other forms of urachal anomalies may require placement of a probe into the urachal tract or instillation of contrast for fistulography. Occasionally, a voiding cystourethrogram will delineate the communication, but is more useful for ruling out other etiologies or associated lower urinary tract anomalies.

D. Treatment. Complete extraperitoneal excision of the urachal malformation is indicated in most cases. Occasionally, infected cysts may benefit from simple drainage and antibiotics; however, most require removal eventually because of the high likelihood of recurrence. Although rare, adenocarcinoma of the urachus has been reported to arise in patients with a history of infected cysts.

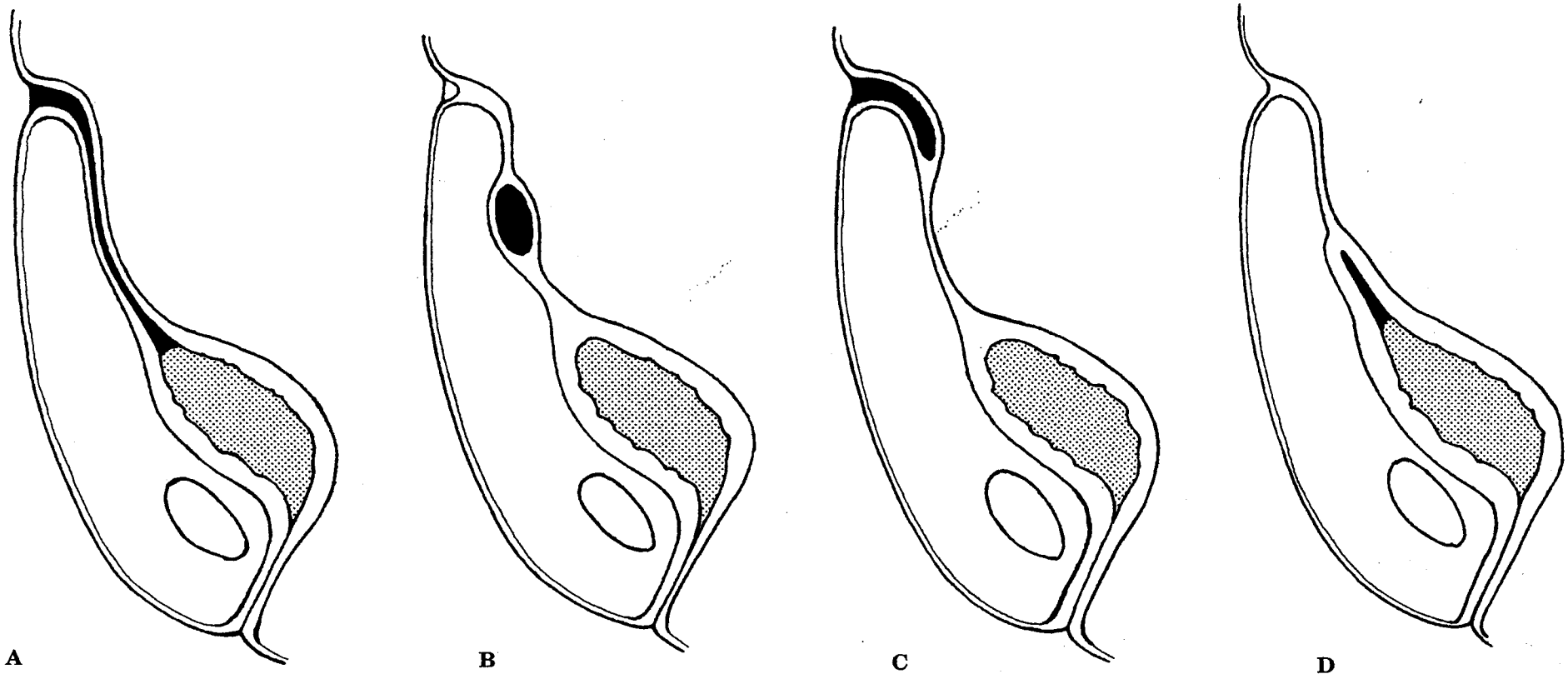


Fig. 16–1. Urachal anomalies. A: Patent urachus. B: Urachal cyst. C: Umbilical sinus. D: Vesical diverticulum.

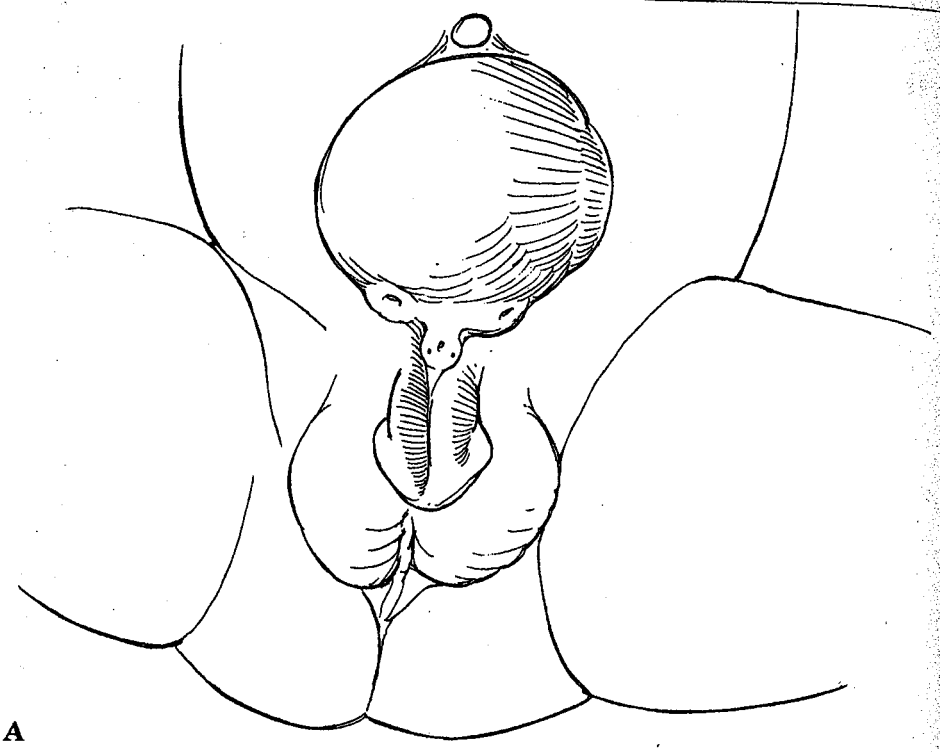
II. EXSTROPHY-EPISPADIAS COMPLEX (Fig. 16–2)

A. Background. This anomaly of the bladder and urethra includes a range of abnormalities from minor epispadias to cloacal exstrophy. Occurring in between 1 and 30,000 to 40,000, it affects boys three to four times more often. Within this spectrum of anomalies, classic bladder exstrophy occurs in 60%, epispadias (all forms) in 30%, and complex cloacal exstrophy and other variants in 10%.

B. Embryogenesis. Persistence of the cloacal membrane after the 4th week of gestation prevents the lateral mesoderm from migrating medially. Once the membrane disappears by the 9th week, the posterior wall of the bladder is exposed to the outside with the umbilicus adjacent to the bladder wall. There is also failure of urethral folding dorsally that causes the epispadiac appearance. The most severe form, cloacal exstrophy, occurs when an abnormally large cloacal membrane perforates prior to division of the cloaca itself by the urorectal septum. When this occurs, an exstrophied bladder separated by an exstrophied ileocecal bowel area results.

C. Anatomy. At birth, the exposed bladder and urethra are obvious malformations (Fig. 16–2A). The bladder plate may vary in size from a small vestigial structure to 6 to 7 cm in diameter. Pubic separation is present in all cases. The upper urinary tracts are generally normal except in cloacal exstrophy patients in whom up to 66% may have abnormalities. Exposure of the bladder results in bacterial colonization, thickening of the bladder with squamous metaplasia, and resultant fibrosis. Once the bladder has been closed, vesicoureteral reflux occurs in the vast majority of patients due to the abnormal placement of the ureters and lack of muscular backing by the bladder. Inguinal hernias are common (both direct and indirect), especially in boys. The testes are descended normally in most cases. Some children may have rectal prolapse (10% to 20%) due to weakness in the perineal floor and the anterior aspect of the levator muscle complex. Adequate bladder closure and approximation of the symphysis may help prevent this.

In girls, the clitoris is divided on both sides of the urethra; the vagina is tilted anteriorly and may be stenotic. There is a higher incidence of müllerian abnormalities such as duplication. Despite this, girls are potentially fertile. In boys, the penis usually is significantly curved upward and shortened. The corporal bodies of the penis are separate (unlike the normal penis in which the corpora communicate) and diverge to attach to the inferior pubic rami that are rotated laterally and anteriorly. Thus, osteotomy performed to restore normal symphyseal distance results in further shortening of the already stubby penis.



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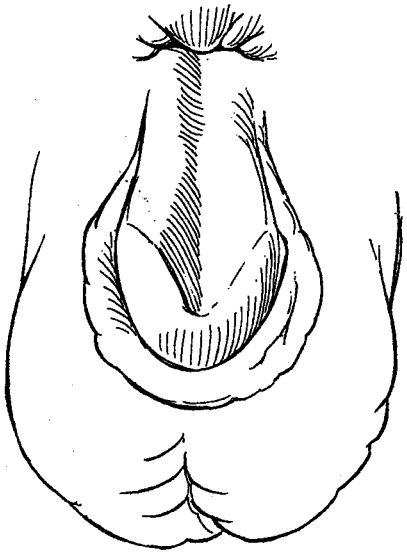

B

Fig. 16–2. A: Bladder exstrophy. B: Epispadias.

D. Initial Evaluation of the Newborn

1. Although upper urinary tract anomalies in classic exstrophy are rare, abdominal ultrasound is important as a baseline. This is especially true for cloacal exstrophy patients.
2. To prevent injury to the bladder, it should be covered with silastic or smooth plastic (e.g., Saran wrap) to avoid contact with diapers and clothing. Ideally, a silk suture should be used to ligate the umbilical stump rather than a plastic or metal clamp to prevent scraping of the bladder.

E. Treatment

1. **Classic bladder exstrophy.** In the distant past, urinary diversion was usually performed by detaching the ureters from the bladder and attaching them to the sigmoid colon (ureterosigmoidostomy). This technique was popular because the presence of an intact rectal sphincter mechanism rendered the children continent. Unfortunately, it became apparent that there was an increased risk for tumor formation at the site of the ureterosigmoid anastomosis and there is a high rate of pyelonephritis with renal loss over time.

   Presently, patients are closed either in a staged approach or most recently, with primary complete bladder and epispadias repair. Primary complete closure or a staged approach provide adequate drainage of kidneys and may enhance the potential for continence after reconstruction.

   a. **Primary bladder and epispadias closure.** In the newborn period, the bladder and epispadias are closed in a single operation. After the immediate newborn period, the pelvic bones may be inflexible and osteotomies are required to bring together the pubic bones. The goal of complete primary closure is to add urethral resistance to the newly closed bladder that should result in bladder cycling and thereby increase bladder capacity and improve long-term continence results. Long-term outcomes determine whether this approach is justified compared to the staged repair.

   b. **Staged approach to bladder exstrophy and epispadias**

      (1) **Primary bladder closure.** Performed during the newborn period, this effectively converts the exstrophied bladder to an epispadias. Some feel that closure in the newborn does not necessitate iliac osteotomies to bring the divergent pubic rami together because of the pliability of the innominant bones and the sacroiliac joint. Others recommend osteotomy strongly, claiming that it contributes to continence in the future by making

bladder and abdominal wall closure tension free (thus less prone to dehiscence), placing the urethra within the pelvic ring, and reapproximating the urogenital diaphragm to enhance voluntary urinary control.

(2) **Epispadias closure** (Fig. 16–2B) is performed between 6 months and 1 year of age. Repair involves correction of intrinsic dorsal curvature (chordee) of the corporal bodies followed by urethral reconstruction that incorporates the same principles utilized in hypospadias repair. Modern repairs bring the urethra from its dorsal position to a more normal ventral position on the penis.

(3) **Bladder neck reconstruction and ureteral reimplantation.** The next stage involves bladder neck tightening along with bilateral ureteral reimplantation to correct reflux. Generally this is performed at 3 to 5 years of age. Bladder capacity at this stage is a significant prognostic feature for future continence. Reconstructing the bladder neck should result in a raised pressure at which urine leaks (called the *opening* or *leak point* pressure, which should be about 30 cm H_2O). Sometimes a suspension procedure of the bladder neck is done to increase the opening pressure.

(4) **Primary epispadias** (Fig. 16–2B). The most common form is that of a proximal urethral opening (penopubic), but other variations may exist including a very mild form of dorsal glanular epispadias. Generally, the more proximal the meatus, the more likely the chance for maldevelopment of the external sphincter and bladder neck continence mechanism. Repair is performed as mentioned for epispadias after bladder exstrophy closure and can be done in the first year of life. Addressing the surgical correction of incontinence should wait until the child is 3 to 5 years old.

(5) **Cloacal exstrophy.** In the past, patients with this most severe and rare form (incidence of 1 per 200,000 live births) of the exstrophy–epispadias complex were usually left to die. Significant problems associated with this include omphalocele, numerous gastrointestinal anomalies (including malrotation, duplication,

duodenal atresia, and Meckel diverticulum), and significant genitourinary (GU) anomalies (including separate bladder halves and bifid genitalia). Newborn boys were occasionally gender converted to female as a result of inadequate genital development and the poor prognosis for developing a normal male phenotype. With modern surgical techniques and a multidisciplinary approach to their care, children with this complex disorder can achieve acceptable lifestyles. The advisability of gender conversion has also been called into question.

F. Prognosis

1. **Continence.** With the current staged approach, continence rates of up to 50% and 60% have been reported, although this varies among treatment centers and surgeons. Many factors influence the outcome, including experience of surgeon, capacity and compliance of the bladder, and the expectations of the family. Continence may improve in boys in adolescence when growth of the prostate occurs. In the majority of patients who remain incontinent after the staged procedures, limited bladder capacity is the cause. Surgical bladder augmentation or creation of a bladder reservoir using bowel can be performed to solve this problem. In a minority of patients the bladder outlet is weak, and placement of an artificial urinary sphincter or urethral augmentation with collagen or Teflon injection may be beneficial.
2. **Renal Function.** Renal deterioration is prevented if bladder pressures remain relatively low and if the child remains on antibacterial prophylaxis because of reflux. In children with more severe forms of exstrophy who may require urinary diversion, upper tract damage can occur and varies according to the type of diversion: 82% for ileal conduits, 22% for nonrefluxing colonic conduits, and 33% for ureterosigmoidostomies.
3. **Obstetric complications**. A common complication in girls with exstrophy is uterine prolapse during pregnancy and delivery. This is thought to be due to a weakened pelvic floor, an abnormally shortened vagina, and failure of development of the cardinal ligaments. Patients who have had prior bladder neck reconstruction or ureterosigmoidostomy should be delivered by cesarean section; those who have had urinary diversion to a stoma should be delivered vaginally if possible to avoid inadvertent injury to the urinary reservoir or conduit. Vaginal delivery is associated with a higher risk of incontinence.

III. IMPERFORATE ANUS (Fig. 16–3)

A. Background. Imperforate anus comprises a spectrum of anorectal anomalies ranging from simple anal fistula to complex cloacal malformations involving multiple organ systems.

1. **Associated anomalies** are seen in up to 50% of cases and urologic involvement in 26% to 50%. Generally, the more severe the defect (i.e., the higher the rectourethral or rectovaginal communication), the higher the incidence of associated anomalies.
   a. **Spinal malformations** include hemivertebra, sacral agenesis or deformation, and spinal dysraphism (spina bifida).
   b. **VACTERL syndrome.** Previously known as VATER (*v*ertebral, *a*norectal, *t*racheo*e*sophageal fistula, *r*adial abnormalities), the terminology has been updated to include *c*ardiac, *r*enal, and *l*imb anomalies.
   c. **GU anomalies.** Major malformations include renal agenesis (20%), vesicoureteral reflux (23%), hydroureter (12%), neurogenic bladder (6%), cystic or dysplastic kidney (5%), renal ectopia (5%), ureteral ectopia (4%), and ureteropelvic junction obstruction (3%). Minor malformations include cryptorchidism (4%), ureteral duplication (3%), hypospadias (3%), and renal malrotation (2%).
2. **Embryology.** By the 8th week of gestation, the primitive hindgut becomes separated into the rectum and the urogenital sinus by the descending urorectal septum. Incomplete migration of the septum with persistence of the cloacal membrane may result in communication (fistula) between the rectum and the urinary tract as well as malformations of the pelvic musculature (sphincter).
3. **Classification.** Anorectal anomalies can be divided into high, intermediate, or low defects based on the level of the fistula between the rectum and the urinary tract or the level of the rectal agenesis (Fig. 16–2).
4. **Morbidity.** Factors contributing to morbidity in these patients relate mainly to GU anomalies, spinal (especially sacral) anomalies, other malformations, and the quality of bowel and urinary sphincters. GU factors include sepsis, absorptive hyperchloremic metabolic acidosis (from absorbed urinary electrolytes via the bowel mucosa), progressive pyelonephritic scarring exacerbated by vesicoureteral reflux, neurovesical bladder dysfunction, and urethral obstruction.

B. Prenatal diagnosis. Although a subtle finding on ultrasound, a dilated distal bowel segment can be a sign. Occasionally, intraluminal calcification is present, which occurs as a result of mixed stagnant urine and meconium.

Fig. 16-3. Imperforate anus. Possible location of rectal genital fistula: (A) boys; (B) girls.

C. **Diagnosis and Management**

1. **Perineal inspection.** The initial evaluation of the perineum yields much information about the severity of the defect and the type of intervention necessary for the infant. In boys, addition of a urinalysis to assess for meconium determines whether a colostomy is needed. If there is any question regarding the diagnosis, a cross-table lateral plain x-ray with the patient in the head down position and with skin marker at the level of the anus (or where it should be), should be done (*invertogram*).
2. **Surgical management.** Most patients with high lesions require a colostomy to divert the fecal stream temporarily until a more distal repair can be accomplished. Distal repair is most often done using the posterior sagittal approach for the anorectoplasty and repair of the rectourinary fistula. The posterior approach is advantageous in preserving the sphincteric musculature (puborectalis sling portion of the levator muscles). In lower or more minor lesions, a posterior anorectoplasty may accomplish the job without having to resort to a colostomy.
3. **Urodynamic evaluation** (see Chapter 11). This is an important part of management because of the possibility of spinal dysraphism or sacral agenesis, and operative injury to bladder innervation.
4. **Urologic interventions**
   a. **Antibiotic prophylaxis.** Until lower urinary tract anomalies are ruled out or reconstructed, all patients should be placed on prophylactic antibiotics to avoid urosepsis.
   b. **Catheterization difficulties.** Often, fistula repair results in urethral narrowing, scarring, or possibly the presence of a diverticulum, all of which make catheterization a challenge. Use of a curved-tip (coudé) catheter may be helpful. If a patient is unable to negotiate the urethra with any catheter and requires intermittent catheterization, placement of a continent catheterizable channel to the bladder (Mitrofanoff appendicovesicostomy) may be a suitable alternative.
   c. **Augmentation cystoplasty.** If bowel segments are necessary to enlarge the capacity of the bladder, avoid using the sigmoid colon so as not devascularize the distal bowel. Also, the ileocecal valve should be preserved to avoid exacerbating stool incontinence as a result of marginal sphincteric function.

IV. PRUNE-BELLY SYNDROME (Fig. 16–4)

A. **Background.** Referred to as Eagle-Barrett, triad, or mesenchymal dysplasia syndrome, all share three major pathologic anomalies: a deficiency, or absence of, abdominal wall musculature; a variety of ureteral,

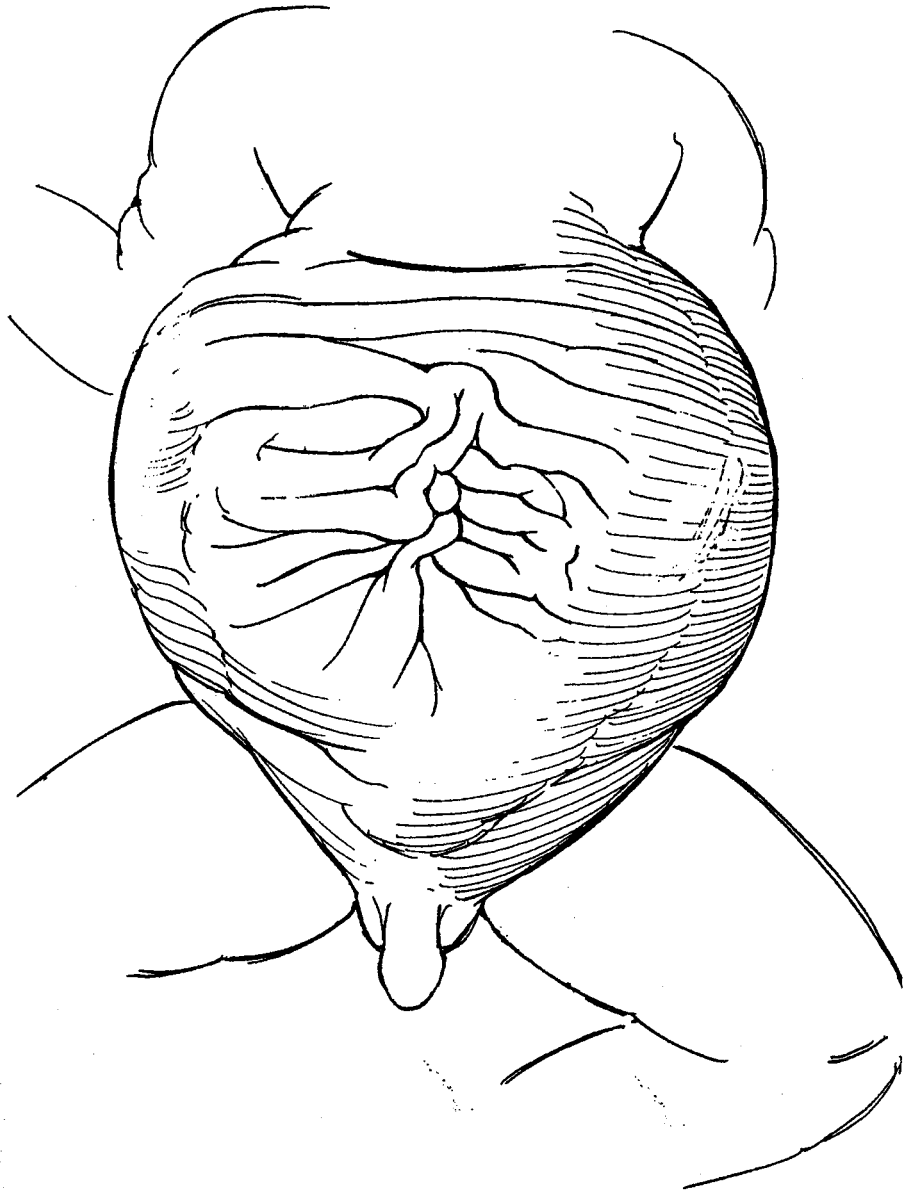

Fig. 16–4. Prune-belly syndrome.

bladder, and urethral anomalies, manifested in most cases by marked dilation; and bilateral undescended testes. Other coexisting orthopedic, pulmonary, and cardiac anomalies have been noted as well. The incidence ranges from 1 in 35,000 to 1 in 50,000 live births with most cases occurring in boys (3% to 5% of cases are in girls).

B. Pathogenesis. The definitive origin of this syndrome remains controversial, but two theories predominate.

1. **Obstructive theory.** This suggests that severe bladder outlet obstruction existed early in gestation and was subsequently relieved after irreversible damage had occurred. This obstruction resulted in bladder distention, ureteral dilation, hydronephrosis, and atrophy of the abdominal wall muscles by the increased pressure, mechanical

distension, and interference with the blood supply of the organs involved. Most patients with prune-belly syndrome lack anatomic obstruction at the time of birth, however.

2. **Mesodermal defect theory.** Because obstruction is rarely present at birth and there is a lack of bladder hypertrophy and hyperplasia, a second theory suggests that a primary defect in mesenchymal development occurs early in gestation.

C. Clinical Manifestations

1. **Kidney.** Renal abnormalities are the major determinants of survival, with a 20% chance of stillbirth or death in the neonatal period from renal dysplasia and the associated pulmonary hypoplasia. An additional 30% of patients develop urosepsis, renal failure, or both in the first 2 years of life.
2. **Ureter.** Severely dilated, and tortuous, the ureters are most severely affected at the lower end and appear histologically to have patchy areas of fibrosis. Vesicoureteral reflux is present in 75% of these patients. Although their radiographic appearance is alarming, drainage is generally adequate.
3. **Bladder.** The bladder is generally capacious, smooth walled, and irregularly thick, without trabeculation. Often a urachal remnant or diverticulum creates an hourglass configuration. Functionally, patients with these bladders exhibit a diminished sensation of fullness and have a large capacity with poor contractility, decreased voiding pressures, and thus a poor ability to empty.
4. **Prostate and posterior urethra.** The prostatic urethra is elongated and characteristically tapers to the membranous region, which gives rise to the typical radiographic appearance of a triangular posterior urethra.
5. **Anterior urethra.** Although the urethra is most often normal in prune-belly syndrome, both urethral atresia and megalourethra can be seen.
6. **Testicles.** Cryptorchidism is seen universally in boys with prune-belly syndrome, the gonads characteristically being found high in the abdomen. Because the gonads are intraabdominal and may be injured further in bringing them down and in addition, the prostate is underdeveloped and the bladder neck open, infertility is the rule. However, in recent years, pregnancies have been achieved using intracytoplasmic sperm injection after retrieval of the occasional sperm.
7. **Abdominal musculature.** The most characteristic manifestation of the syndrome is the wrinkled, prunelike skin of the abdomen in the newborn infant. Skeletal muscle hypoplasia is seen in all three layers of the abdominal wall muscles. Complications are surprisingly minimal. The inability to sit up directly from the supine position may delay the

onset of walking, but it rarely affects normal physical activity.

8. **Other associated anomalies.** Other anomalies are found in over 65% of patients, the most common being cardiopulmonary and gastrointestinal, and including orthopedic and developmental problems. Pulmonary abnormalities range in severity, with the most significant arising in cases with associated antenatal oligohydramnios. Children who survive the neonatal period usually have no associated pulmonary problems.

D. Fetal diagnosis and treatment. Antenatal ultrasound is capable of detecting abnormal urinary tract dilation as early as 14 weeks. However, it is difficult to differentiate prune-belly syndrome from other causes of urinary tract dilation. In the presence of oligohydramnios, prenatal intervention has been advocated to decompress the dilated bladders and restore the amniotic fluid volume. However, although prenatal intervention may improve pulmonary function, its effectiveness in improving renal function is uncertain. Furthermore, intervention in cases of prune-belly syndrome is difficult to justify because in utero obstruction may not exist.

E. Management of the Neonate

1. **Immediate evaluation**
   a. **History and physical.** Rule out life-threatening cardiac and pulmonary problems first. The abdominal examination is made much easier by the thin, relaxed belly, which permits easy palpation of the intraabdominal and retroperitoneal contents.
   b. **Serum creatinine** levels in the first days of life reflect maternal values, but a progressive increase (or failure to decline) suggests some degree of renal insufficiency.
   c. **Bladder stimulation test.** Simple bladder massage can be used to stimulate a detrusor response, and one can use this as a test of voiding.
   d. **Imaging**
      (1) Begin with an ultrasound that can provide information on renal potential and bladder emptying.
      (2) Voiding cystourethrography (VCUG) should be postponed to avoid introducing bacteria into a stagnant system. However, if renal function is abnormal, a VCUG should be done early. Aggressive antibiotic treatment is essential around the time of VCUG as the introduction of even a few bacteria can result in sepsis in the stagnant urinary tract.
      (3) A dimercaptosuccinic acid scan may be done to evaluate for renal malformation and scarring.

2. **Treatment.** Management is decided based on whether patients are severely affected (oligohydramnios, pulmonary hypoplasia, or pneumothorax); moderately affected with typical external features and uropathy of the full-blown syndrome, but without immediate problems with survival; or have a mild form unlikely to develop urosepsis or azotemia. The third category of patients show internal features that may be mild or incomplete. Uropathy is less severe and renal function is stable.
   a. **Severely affected** patients in the first category usually do not survive the neonatal period; however, for the few who do survive, urinary diversion by vesicostomy, cutaneous pyelostomy, or ureterostomy is often recommended to provide optimal urinary drainage.
   b. **Moderately affected.** The approach to the second category of patients with prune-belly syndrome is variable, but is usually nonoperative unless infection (despite antibiotic prophylaxis), failure of adequate renal growth, or decreasing renal function is encountered. In these cases the urinary tract is reconstructed with ureteral tailoring and correction of reflux to reduce stasis. Reconstruction of the abdominal wall as well as orchidopexy are often performed simultaneously.
   c. **Mildly affected** patients in category three who have good renal function do not usually require surgical intervention. However, lifelong antimicrobial prophylaxis should be instituted (in neonates, amoxicillin or first-generation cephalosporin; later, trimethoprim-sulfamethoxazole or nitrofurantoin). Orchidopexy may be delayed until such time as any other reconstructive procedures may become necessary or at about 6 months.

F. **Management into childhood and beyond.** Abnormalities of bladder drainage are the principal source of problems that may lead to renal deterioration.
   1. **Evaluation: urodynamics.** The child with a low urinary flow rate and a significant residual volume should be evaluated with urodynamics (see Chapter 12).
   2. **Treatment**
      a. In some cases, endoscopic evaluation of the urethra with internal urethrotomy of pseudovalves may be attempted to reduce the outlet resistance, although this remains controversial.
      b. **Ureteral surgery.** Although pyelonephritis or renal deterioration may prompt one to reimplant and perhaps tailor the ureters to improve drainage and prevent reflux, these procedures are complicated by the poor peristaltic nature

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of these ureters and the abnormal bladder into which they are reimplanted. Although the radiographic appearance of the urinary tract improves after surgery, it remains to be seen whether there is any long-term functional improvement. Because of persistent stasis postoperatively, nearly all these patients are maintained on lifelong antibacterial prophylaxis.

c. **Clean intermittent catheterization.** Adequate drainage can be obtained with clean intermittent catheterization, but this can be difficult because these patients have normal urethral sensation and some have urethral anomalies. For this reason, a continent abdominal stoma may be useful. If the family cannot catheterize the patient, a urinary diversion, usually a cutaneous ureterostomy, can be life saving until the child is mature enough to perform the catheterization.

3. **Prognosis**

a. **Renal function.** Patients who survive infancy with mildly impaired renal function may develop renal failure as a result of chronic pyelonephritis and reflux nephropathy. In these individuals, renal transplantation can be performed successfully. All these patients should perform clean intermittent self-catheterization to alleviate chronic retention.

b. **Testicles.** Early intraabdominal orchidopexy is warranted in boys with prune-belly syndrome because repair in infancy allows placement of the testes into the scrotum without division of the spermatic vessels, which may not be possible later in life. Although likely to be infertile, these patients may benefit from advances in fertility techniques. In addition because these patients have the same risk for testicular malignancy as normal boys with undescended testes, having gonads in the scrotum will facilitate examination.

c. **Abdominal wall.** Major reconstruction of the abdominal wall repair can be done at the same time as orchiopexy. This procedure includes excision of redundant prunelike folds of skin and fascia to create a satisfactory waistline. In milder cases, observation may be best because of the tendency for the abnormal abdominal wall to stretch out again.

RECOMMENDED READING

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