GENITOURINARY PROBLEMS IN PEDIATRICS

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Foreword

Pediatricians and other physicians who treat children frequently encounter conditions that require a urologic evaluation. Because many of these conditions are unique to infants and children, they are managed best by urologists who devote their time entirely to urologic diseases of childhood. There is now a growing number of pediatric urologists who by concentrating on children have contributed a valuable body of knowledge and have been responsible for many of the diagnostic and therapeutic advances in recent years.

Two well-known leaders in this field have authored this monograph. Dr. George Kaplan received his medical degree and urologic training at Northwestern University. He then joined the faculty of the University of California, San Diego, where he is currently Clinical Professor of Surgery and Chief of Pediatric Urology. He is a contributing author of more than 80 papers. Dr. A. Barry Belman also obtained his medical and urologic training from Northwestern. He is at present Professor of Urology and Child Health at George Washington University and Chairman of the Department of Pediatric Urology at Children's Hospital National Medical Center in Washington, D.C. Dr. Belman has authored or coauthored more than 70 papers and books.

This book was written primarily for the generalist who cares for children. It is clinical in orientation and contains a great deal of practical information on problems ranging from, in the words of the authors, "the common—bedwetting—to the rare—exstrophy of the bladder—and from the simple—cystitis—to the overwhelmingly complex—myelomeningocele."

MILTON MARKOWITZ, M.D.

Preface

The publication of this volume as part of a series dedicated exclusively to pediatric medicine emphasizes the importance pediatric urology has achieved as a discipline over the past two decades. Largely owing to the devotion of a relatively few physicians with great foresight, it became evident in the early sixties that the urologic problems seen in children required a unique perspective and, with that perspective, a separate approach from that employed with adults. During that same period advances in pediatric imaging, anesthesia, and nephrology, as well as improved optics and miniaturization of endoscopic instruments, transformed the field. With centralization of care in major children's hospitals, the opportunity then existed for those few individuals who were devoting their efforts primarily or exclusively to the care of the genitourinary systems in children to study the approaches and treatment of problems such as urinary tract infection, vesicoureteral reflux, the obstructed urinary tract, genital abnormalities, and other major anomalies. Through these combined labors great advances were made.

Over the ensuing years a consensus has developed regarding the approach to many genitourinary problems of childhood, although by no means are all questions resolved, nor is there agreement over all approaches to diagnosis or therapy. In this volume we have attempted to present an overview of problems of the genitourinary tract that the primary physician might face in pediatric care. In this endeavor we have leaned heavily upon the experience of others and are grateful to Drs. Panayotis P. Kelalis and Lowell R. King for allowing us to use numerous illustrations from Clinical Pediatric Urology (W. B. Saunders Co., 1976) and to Drs. J. Hartwell Harrison and Alan Perlmutter for permitting the use of illustrations from the pediatric portion of the fourth edition of Campbell's Urology (W. B. Saunders Co., 1979).

We have relied upon our own clinical experience in many areas; however, particular effort has been made to provide references for problems that remain controversial. While definitive statements can be made in reference to many of the subjects addressed, significant topics of concern remain that cannot be conclusively addressed at present. These include the question of surgical versus medical management of vesicoureteral reflux, which is currently being evaluated prospectively under an international protocol. The optimal age for genital surgery is as yet unknown and awaits definitive study to ascertain the psychological and functional implications. Although great strides have been made in the treatment of certain neoplasms through national and international study groups, there are other areas in which little progress has been made. Progress can only result from pooling our limited

resources, especially in instances of rarer tumors. By maintaining open communication, we can continue to improve our store of knowledge and thereby follow the example of those who have led us into this most gratifying subspecialty.

Finally, we wish to offer our appreciation to the numerous individuals who were supportive of this endeavor: Mrs. Hilary Kavanagh, Mrs. Mary Moyta, and Ms. Bette Jo Garrett for exhaustive secretarial help and Mrs. Deborah Gilbert for her untiring assistance in locating references. We are particularly grateful to Mr. Jack Hanley of W. B. Saunders Co., who originally nurtured the idea for this work, and to Mary Cowell and her associates for their editorial assistance and efforts in gathering the various borrowed illustrations.

Contents

Chapter One

UROPATHOLOGY IN THE PEDIATRIC PATIENT

Pediatric urology is a discipline that encompasses a spectrum of problems ranging from the common — bedwetting — to the rare — exstrophy, and from the simple — cystitis — to the overwhelmingly complex — myelomeningocele. Congenital anomalies of the urogenital tract affect a signficant percentage of the population. Although some are inconsequential, others result in major morbidity and occasionally mortality. Additionally, urinary tract infection is one of the commonest bacterial diseases seen in the pediatric population. Despite the frequency of uropathologic conditions in children, pediatric urology is only now maturing as a separate discipline. Consequently, "the state of the art" and the progress made in this subspecialty is often poorly appreciated.

The diagnostic capabilities for evaluation of the urinary tract have recently made tremendous advances. It is, therefore, usually not difficult to arrive at the proper diagnosis of a urologic problem if one only suspects that such a problem exists. Most acute as well as the more chronic problems of the urinary tract will be associated with some symptom or readily documented sign that will lead the thoughtful observer to a correct diagnosis. Even infants with urinary tract infections usually exhibit some change in urinary habits; if not, some other sign, such as fever or failure to thrive, will become evident. It is truly unfortunate when these symptoms remain unrecognized for a protracted period of time prior to evaluation, as most serious problems of this system are amenable to treatment if the diagnosis is made promptly.

It is a truism that before being able to arrive at a diagnosis, one must first consider the existence of a disease as a possibility. This statement is particularly applicable to disorders of the urinary tract since significant information can often be obtained with simple and straightforward studies, such as history, physical examination, urinalysis, and urine culture. Because it is often a little more difficult to obtain a urine specimen in the population in question, routine urinalysis has not achieved the prominence in pediatric practice that it has in adults. With just a modicum of forethought and patience, specimens can be collected routinely, even in infants. Toilet-trained children, when reassured, will usually void on request, and infants urinate frequently when well hydrated. When the specimen obtained is collected in a sterile container, it then can also be used for culture. Urine cultures have not been used routinely as they are perceived as being

costly. The recent introduction of "minicultures" has removed this objection. Obviously, any positive culture obtained from a voided, bagged collection in an infant should be confirmed by a second, fresh specimen or, preferably, by suprapubic aspiration or catheterization prior to treatment or radiographic evaluation.

The question often arises as to the benefit of routine urine screening in children. The term "screening" is generally applied to public health programs designed to cover large population groups, but in this context we will consider it only as it relates to well-child care. If one assumes that all infants and children receive some regular pediatric care, then sometime prior to school age, urine screening cultures are indicated. This applies most specifically to girls.

The incidence of urinary tract infection in school-age girls has been tabulated at between two and five per cent. Girls are affected 20 times more frequently than boys. Over 15 per cent of children with asymptomatic bacteriuria have radiographic changes consistent with bacterial pyelonephritis (renal scarring), and one third have vesicoureteral reflux.

The incidence of pyelographic "renal scars" in adolescents and adults is the same as that seen in children. This implies that if these findings are acquired and are secondary to infection, the causative episode was overlooked. Even in those children discovered to have bacteriuria before one year of age, pyelonephritic renal scarring was observed in one third of these cases. It is becoming increasingly apparent that the renal damage associated with vesicoureteral reflux and bacteriuria has its onset in infancy.

Urine cultures performed periodically at six-month intervals for the first two or three years of life and then annually may be the only way renal scarring can be minimized in this otherwise asymptomatic group. This recommendation assumes that (1) abnormalities detected are promptly treated, (2) that early treatment can prevent these changes, and (3) that the radiographic findings noted are not congenital but do actually result from acquired bacterial infection.

PRESENTING SYMPTOMS

Voiding symptoms, the most common and easily recognizable presenting urologic complaints, are directly referable to the urinary tract. Urinary urgency, incontinence, and dysuria are indicative of acute lower urinary tract inflammation. Pyuria also suggests an inflammatory process. Nonetheless, these symptoms, even when coupled with the finding of pyuria upon microscopy of the urinary sediment, are insufficient evidence for diagnosis of urinary tract infection.

As will become clearer in subsequent chapters, the diagnosis of urinary tract infection carries with it a responsibility to prove that the affected child does not harbor an underlying anatomic abnormality that contributed to that infection. Hence, all children other than black girls with documented urinary tract infection must, in our opinion, be studied at least by cystography and excretory urography. Radiographic evaluation is invasive, expensive, and time-consuming and should not be performed indiscriminately. However, evaluation is essential in children with proven urinary tract infection. Black girls have a significantly lower incidence of anatomic urinary abnormalities and therefore represent an exception to the recommendation for radiographic evaluation of bacteriuria responsive to treatment and not associated with fever (Chap. 4).

INCONTINENCE

One of the more frequent presenting symptoms is the involuntary loss of urine. To better understand this symptom and its etiology, several parameters must be detailed. It should be determined whether the enuresis is primary (present since birth) or of secondary onset (developing after an interval of normal control), whether it is nocturnal, diurnal, or both, and whether the child dribbles continuously or intermittent-

ly. In addition, it is important to learn whether there also are irritative symptoms, such as urgency or dysuria, and how often the child voids. The force and character of the urinary stream, as well as discovering if the child has periods of normal control, are significant.

Urinary incontinence may exhibit a certain pattern, and this will often lead to the underlying problem. Total incontinence is the inability to store any urine and indicates that the urinary sphincters are anatomically or functionally absent (as in patients with epispadias or myelomeningocele) or that they have been bypassed (such as in vesicovaginal fistula). Overflow incontinence occurs when the urinary outlet is obstructed and the patient dribbles frequently to relieve a consistently full bladder. Urge incontinence occurs when there is a sudden and uncontrollable need to void that cannot be suppressed. This implies bladder irritability. Precipitate voiding is much the same but occurs without a preceding urge to void and suggests neurologic origins. Stress incontinence occurs when intravesical pressure momentarily exceeds infravesical resistance (such as in "giggle incontinence"). Lastly, paradoxic incontinence describes the girl who is always wet and yet voids normally. This is suggestive of an ectopic ureteral orifice outside the urinary sphincter mechanism.

IRRITATIVE SYMPTOMS

Irritative symptoms, such as dysuria, frequency, and urgency, are indicative of bladder hyperactivity. Children so affected often have urinary incontinence with urgency. This is usually secondary to inflammation and may also be associated with suprapubic or perineal pain.

HEMATURIA

Hematuria is a common problem for which children are referred to the urologist or nephrologist. Since hematuria is a symptom shared by many diverse disorders (Table 1–1), it is advantageous to perform some preliminary screening studies prior to for such preliminary evaluation is found in Table 1–2.

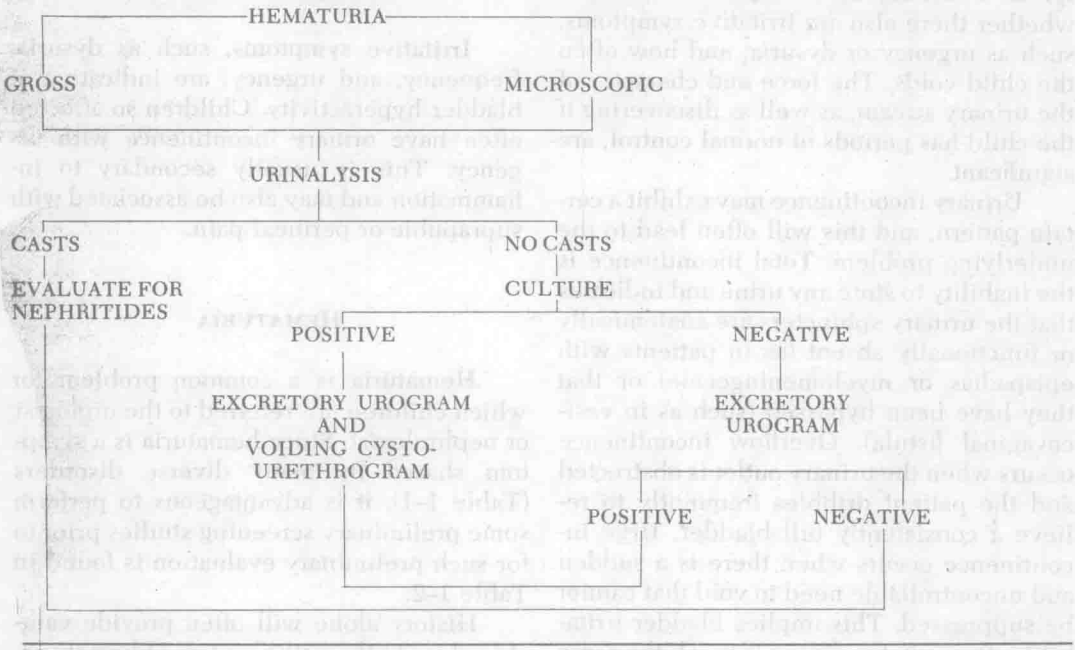
History alone will often provide valuable clues to the pathogenesis of hematuria. Total hematuria, i.e., blood throughout the deciding the propriety of referral. A schema urinary stream, is of renal or bladder origin. Patients with urinary urgency, frequency, and dysuria along with hematuria are likely to have hemorrhagic cystitis. Urine culture will corroborate the diagnosis in most cases of bacterial infection. Confusion can result in children with viral cystitis, in which case routine bacterial cultures will be negative.

Table 1–1. Causes of Hematuria

MEDICAL	SURGICAL
Urinary tract infection	Urinary tract infection
Acute glomerulonephritis	Obstructive uropathy
Rapidly progressive glomerulonephritis	Trauma
Chronic hypocomplementemic glomerulonephritis	Neoplasm
Henoch-Schöenlein nephritis	Vascular malformation
Lupus erythematosus nephritis	congenital (AV fistula, hemangioma)
Goodpasture's syndrome	acquired (AV fistula, renal vein thrombosis)
Hemolytic uremic syndrome	Foreign body
Benign hematuria	
Familial hematuria	
Periarteritis nodosa	
Subacute bacterial endocarditis	
Sepsis	
Bleeding diathesis	
Drug toxicity	
Urethral meatal excoriation	

(Modified from Burke, R. H.: Urinalysis; The investigation of hematuria and renal biopsy. *In* Kelalis, P. P., King, L. R., and Belman, A. B.: Clinical Pediatric Urology, Vol. 1. Philadelphia, W. B. Saunders Co., 1976.)

Table 1–2. Schema for Evaluation of Pediatric Hematuria



Initial or terminal hematuria suggests that the responsible lesion is at the bladder base or in the proximal urethra. Although radiographic evaluation, including voiding cystourethrography, is recommended, this type of bleeding is poorly understood and rarely is of significance in children. Blood staining of the underwear in boys without true hematuria is almost always of urethral origin and secondary to either a nonspecific and poorly understood urethritis or urethral irritation. In the absence of actual hematuria, evaluation of such problems with studies additional to urine culture and excretory urography with a film during voiding will probably contribute little.

In infant males, ammoniacal meatitis may cause blood staining on diapers. The underlying cause should be obvious, and local treatment is all that is necessary. Bright red urine, especially with clots, suggests urologic pathology, whereas tea-colored or smoky urine is more likely nephrologic in origin. It is to be emphasized that all red urine is not necessarily bloody, and microscopic or chemical confirmation of hematuria is advisable before embarking on further studies. Causes for discoloration of the urine are listed in Table 1–3.

A history of recent upper respiratory tract illness or streptococcal infection suggests post-streptococcal glomerulonephritis, particularly in younger children. Such children, who also have edema, hypertension, and fever, do not constitute a diagnostic dilemma. Those children found to have red blood cell casts on microscopic urinalysis require no further urologic evaluation but should have appropriate studies to elucidate the cause of the nephritis. Appropriate laboratory studies, including a throat culture, serum complement level, and antistreptolysin-O (ASO) titer or streptozyme reactivity, are in order prior to further evaluation in all children presenting with asymptomatic gross or microscopic hematuria.

A history of trauma can often be obtained when dealing with children since their level of activity is so high. However, urinary obstruction with a dilated, tense system proximal to the site of obstruction becomes very susceptible to injury. There-

Table 1–3. Causes of Urinary Discoloration

CAUSES OF ABNORMAL URINE COLOR
Colorless:
Diabetes mellitus, diabetes insipidus, dilution of urine.
Milky:
Chyluria, pyuria.
Yellow:
Normal, phenacetin, quinacrine, riboflavin.
Red:
Hemoglobinuria, myoglobinuria, hematuria, chronic lead and mercury poisoning; eosin, acetophenetidin, diphenylhydantoin, phenindione, emodin, anisindione, phenolphthalein, phenothiazines, phensuximide, rifampin; anthocyanin.
Blue, Blue-green to Green:
Biliverdin, indicanuria; amitriptyline, anthraquinone, arbutin, flavin derivatives, indigo blue, indigo carmine, methylene blue, tetrahydronaphthalene, thymol, phenol, resorcinol, salol, toluidine blue, triamterene.
Orange:
Dehydration; santonin, cryptophanic acid, salicylazosulfapyridine, phenazopyridine.
Brown:
Porphyria, presence of urobilinogen; nitrofurantoin, primaquine, chloroquine, furazolidone, metronidazole, argyrol; fava beans, aloe.
Brown black:
Hemorrhage, melanin, homogentisic acid (alcaptonuria), p-hydroxyphenylpyruvic acid (tyrosinosis); methyldopa, cascara, pyrogallol, iron sorbitol, methocarbamol, senna, phenylhydrazine; rhubarb.
Note: Malingerers sometimes color their urine with blood, food coloring, or colored crepe paper; or by ingesting food or substances that will color it.

(From Elkins, M., and Kabat, H.: Causes of urinary discoloration. Am. J. Hosp. Pharmacol., 25:489–519, 1968.)

fore, hematuria, even when associated with mild trauma, requires radiographic evaluation.

FEVER

Otherwise unexplained fever should direct one's attention to the urinary tract as a possible etiologic source. When fever accompanies urinary tract infection, there is an increased likelihood that one is dealing with bacterial pyelonephritis rather than cystitis. Govan and colleagues (1975) found that 60 per cent of children with febrile urinary tract infection had demonstrable vesicoureteral reflux and, hence, presumed pyelonephritis. Since bacterial pyelonephritis can lead to renal scarring, it is desirable to recognize this problem so that it can be treated as rapidly as possible.

PAIN

Abdominal pain is a very common symptom in children. However, it is rarely due to anatomic pathology of the urinary tract. Pain following high fluid intake may be the result of acute dilatation of the urinary tract behind an obstruction (ureteropelvic junction obstruction). Ultrasonographic screening is a reasonable, noninvasive means to rule out long-standing obstruction as a cause for pain. Occasionally excretory urography must be performed to insure that the urinary tract is not the underlying cause. Lower abdominal discomfort may also be attributable to cystitis. If questioned closely, such children often have a history of squatting and pressing the perineum with a heel at the time of these pains to prevent incontinence. This complex is only seen in girls and is explained by a very irritable bladder usually brought about by chronic cystitis (Chap. 3).

Back pain in children is rarely caused by urinary tract pathology. Musculoskeletal problems are a much more common cause. Children localize abdominal pain poorly. Even when unilateral ureteral obstruction is the cause, the pain is frequently perceived periumbilically. Acute obstruction is more likely to cause pain than is the chronically dilated urinary tract.

Flank pain is an uncommon symptom in children, although children with acute pyelonephritis may localize pain to the ipsilateral kidney. Occasionally, children with severe vesicoureteral reflux complain of flank discomfort associated with voiding that disappears shortly after completion of that act. Acute distension of the upper urinary tract from large volume reflux is the cause.

PHYSICAL EXAMINATION

Inspection of the patient may suggest certain diagnoses. Microcephaly can be

seen in association with obstructive uropathy, and hydrocephalus suggests neurogenic bladder disease (in association with myelomeningocele). The recognition of Potter's facies will lead to a prompt diagnosis of renal agenesis or severe obstruction. Many of the chromosomal abnormalities are also manifested by physical appearance.

Bladder or cloacal exstrophy, myelomeningocele, imperforate anus, and sirenomelia are easily recognizable at birth, and all suggest the need for further urologic assessment. The blueberry muffin syndrome (cutaneous neurofibromata and café au lait spots) are related to various neural crest tumors.

Low-set and malformed ears also suggest renal abnormalities. Webbing of the neck is found in Turner's syndrome. Laterally placed nipples occur in Turner's and Noonan's syndromes. Pneumomediastinum and pneumothorax are occasionally seen in newborn males with obstructive uropathy.

Urologic abnormalities in childhood may also be completely asymptomatic but offer the opportunity to be discovered in the course of routine physical examination. The first such opportunity is the initial newborn examination, but many others present during the child's first several years of life. We have chosen to discuss findings in the order in which they may be discovered rather than in order of frequency or importance. In addition, certain organ system abnormalities or constellations of abnormalities often have associated developmental urinary tract abnormalities. Awareness of such associations will lead the astute clinician to an early diagnosis.

Table 1–4. Differential Diagnosis of Abdominal Masses Referable to the Genitourinary Tract

Hydronephrosis
Ureteropelvic junction obstruction (may be bilateral)
Ureterovesical obstruction (may be bilateral)
Posterior urethral valves (hydronephrosis may be unequal)
Ectopic ureter with obstruction
Ureterocele (almost always associated with duplication)
Cystic dysplasia (multicystic kidney)
Polycystic disease
Infantile (autosomal recessive)
Adult (autosomal dominant)
Wilms' tumor
Mesoblastic nephroma (neonate)
Neuroblastoma
Idiopathic adrenal hemorrhage of newborn
Renal vein thrombosis
Renal ectopia
Horseshoe kidney (palpable isthmus)
Hamartoma (associated with tuberous sclerosis)
Renal cell carcinoma (rare in children)
Retroperitoneal sarcoma or teratoma
Distended bladder
Posterior urethral valves
Neurogenic bladder
Hydrometrocolpos
Lower genitourinary rhabdomyosarcoma

HYPERTENSION

An often neglected part of the child's physical examination is measurement of blood pressure. This is probably a reflection of the low incidence of blood pressure elevation in the pediatric population. Nevertheless, hypertension can be a serious problem in children, and its recognition demands a thorough search for an underlying, reversible cause.

ABDOMINAL EXAMINATION

Careful abdominal palpation, particularly in infants, can be rewarding (Table 1–4). The art of medicine implies a certain skill and patience that separates the novice from the master. This especially applies to

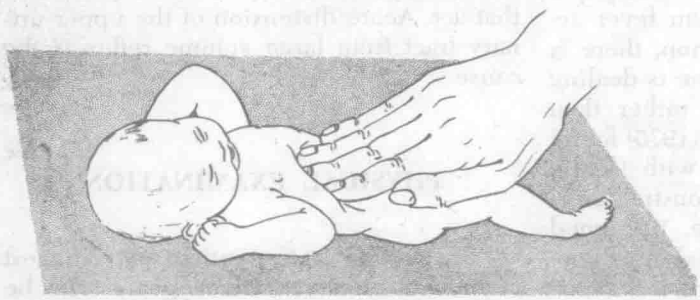


Figure 1–1. Method of abdominal palpation in an infant. Note that the flat portion of the fingers is used rather than the tips. (From Kelalis, P. P., King, L. R., and Belman, A. B.: Clinical Pediatric Urology, Vol. 1. Philadelphia, W. B. Saunders Co., 1976, p. 110.)

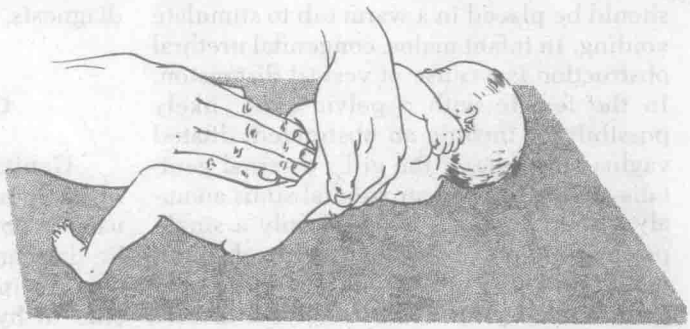


Figure 1–2. Method of left renal palpation in an infant. The examiner's right hand lifts while the left hand explores the left upper quadrant and flank. (From Kelalis, P. P., King, L. R., and Belman, A. B.: Clinical Pediatric Urology, Vol. 1. Philadelphia, W. B. Saunders Co., 1976, p. 111.)

the abdominal examination. Recently we have had occasion to see two patients, one with a Wilms' tumor and another with a neuroblastoma. Early diagnosis resulting in complete resection of a localized lesion was accomplished in each instance because the primary physician in each case refused to allow children to leave his office until an adequate abdominal examination had been performed.

Initially, examination of the abdomen should be carried out with the patient supine, thoroughly palpating all four abdominal quadrants. Ectopic and horseshoe kidneys may be found easily. Abdominal relaxation is usually required to satisfactorily feel normally sized and positioned kidneys and to deteot small renal or suprarenal masses. Such relaxation can often be produced by having the older child breathe in and out deeply while probing, especially during the expiratory phase. In infants and younger children it may be helpful to use a pacifier rather than a bottle. A full stomach often prevents adequate examination. Counterpressure in the flank region aids considerably in "trapping" the kidney after inspiration, allowing for better palpation (Figs. 1–1 to 1–3).

Abdominal masses in the newborn period arise within the urogenital systems in more than two thirds of those with this finding. The incidence of hydronephrosis and multicystic kidneys in infants in this group is approximately equal. Other less common causes include renal vein thrombosis, polycystic kidneys, and solid renal tumors.

The sudden discovery of an abdominal mass, often of mammoth proportions, can be a most startling finding in infants and children. In the toddler and older children, tumors of the kidney, the adrenal, or the sympathetic chain should be suspected. However, unrecognized hydronephrosis is also a definite consideration. Evaluation should be immediately pursued starting with either excretory urography or ultrasonography.

A distended bladder may be the result of voluntary urinary retention brought about by dysuria. It is not uncommon for children to refrain from urinating for 18 hours voluntarily because of pain. Catheterization

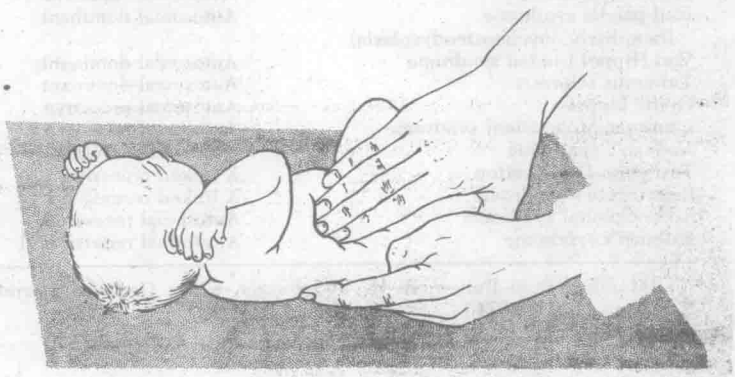


Figure 1–3. Method of right renal palpation in an infant. The examiner's left hand lifts while the right hand explores the right upper quadrant and flank. (From Kelalis, P. P., King, L. R., and Belman, A. B.: Clinical Pediatric Urology, Vol. 1. Philadelphia, W. B. Saunders Co., 1976, p. 111.)

should not be carried out. Instead, the child should be placed in a warm tub to stimulate voiding. In infant males, congenital urethral obstruction is a cause of vesical distension. In the female with a pelvic mass, likely possibilities include an obstructed, dilated vagina or uterus. If the girl's external genitalia are abnormal, a urogenital sinus anomaly should be considered. If only a single perineal opening is present, a cloacal abnormality in which urinary, fecal, and internal genital tracts join is most likely. A mobile lower abdominal mass in a girl may be ovarian in origin.

Frequently, examination of the lower abdomen will reveal what is at first thought to be an abdominal mass but what subsequently is proved to be stool in the colon. A repeat examination following voiding helps in differentiating stool from a distended bladder. However, rectal examination should always be carried out in any child with unexplained abdominal symptoms or findings and will often eliminate this source of diagnostic error. Sarcomas of the lower genitourinary tract are palpable rectally as well as abdominally and hence will not be overlooked if considered in the differential diagnosis.

GENITAL EXAMINATION

Genital abnormalities as a rule are rare in girls. In most males such problems are usually obvious and do not pose a diagnostic dilemma. Hypospadias should be obvious. An incompletely formed prepuce is a clue to hypospadias and can direct one's attention to more mild forms. If hypospadias is identified, circumcision should not be performed until the child has been evaluated by a surgeon accustomed to dealing with this problem frequently. The evaluation of the newborn with ambiguous genitalia is discussed more fully in subsequent chapters. The presence or absence of testes should be documented on initial examination.

When examining the newborn's penis, it is unrealistic to expect to separate the foreskin from the glans without force. This finding should not be termed "phimosis" but rather failure of separation of the tissue

Table 1–5. More Common Hereditary Diseases Due to Single Mutant Genes

DISEASE	GENETIC TRANSMISSION	GU EFFECT
Hereditary nephritis with deafness (Alport's syndrome)	Autosomal dominant	Renal failure
Benign familial hematuria	Autosomal dominant	Hematuria
Adult polycystic disease	Autosomal dominant	Renal cystic disease
Infantile polycystic disease	Autosomal recessive	Renal cystic disease
Medullary cystic disease (juvenile nephronophthisis)	Autosomal recessive	Renal failure
Diabetes insipidus, nephrogenic	X-linked	Polyuria, dilated collecting system
Ehlers-Danlos syndrome	Autosomal dominant	Ureteropelvic junction obstruction
Sickle cell disease	Autosomal recessive	Priapism, hematuria
Sickle cell trait	Autosomal recessive	Hematuria, papillary necrosis
Nail-patella syndrome (hereditary onychoosteodysplasia)	Autosomal dominant	Renal insufficiency hydronephrosis
Von Hippel Lindau syndrome	Autosomal dominant	Wilms' tumor
Tuberous sclerosis	Autosomal dominant	Renal hamartoma and cystic disease
Cystic fibrosis	Autosomal recessive	Atresia of vas deferens
Laurence-Moon-Biedl syndrome	Autosomal recessive	Hypogonadism
Noonan's syndrome	Autosomal dominant	Cryptorchidism, renal anomalies
Testicular feminization	X-linked recessive	Spectrum of genital abnormalities
Reifenstein's syndrome	X-linked recessive	
Adrenogenital syndrome	Autosomal recessive	
Kallman's syndrome	Autosomal recessive	

(Modified from Burger, R. H., and Burger, S. E.: Genetic determinants of urologic disease. Urol. Clin. North Am., *1*:419, 1974.)

Table 1–6. Chromosomal Anomalies

Klinefelter's syndrome	47, XXY	Eunuchoid appearance, small gonads, infertility
Turner's syndrome	45, XO	Infertility, renal anomalies
Mixed gonadal dysgenesis	45, XO-46, XY mosaicism	Ambiguous genitalia, wolffian and mullerian structures present, gonadal neoplasia
XYY syndrome	47, XYY	Cryptorchidism, small penis, hypospadias
XXXXY syndrome	49, XXXXY	Cryptorchidism, small penis
Down's syndrome (Trisomy 21)		Genital abnormalities, horseshoe or cystic kidneys
Trisomy 13		Cystic kidneys, bicornuate uterus, cryptorchidism
18q deletion syndrome		Hypoplastic genitalia

(Modified from Burger, R. H., and Burger, S. E.: Genetic determinants of urologic disease. Urol. Clin. North Am., *1*:419, 1974.)

planes, an entirely normal phenomenon. It is not uncommon to see incomplete separation of these layers lasting until three to five years of age or longer.

Neurologic Examination

Finally, some degree of attention should be paid to the spine and neurologic systems. The most common malformation affecting the innervation of the lower urinary tract is myelomeningocele and is recognizable at birth in almost all affected children. Less dramatic but potentially serious midline closure defects may be suspected by tufts of hair or hemangiomata overlying the lower sacral spine. Physical examination should include evaluation of anal reflexes, sensation, and sphincter tone.

INHERITED DISEASES

Hereditary diseases due to a single mutant gene are determined by simple mendelian laws of inheritance. Those problems are relatively easily categorized and have been well studied. The most common of those that involve the genitourinary tract are listed in Table 1–5.

Chromosomal Abnormalities

There are many problems of urologic interest that are recognized as being secondary to noninherited chromosomal abnormalities. Common examples that involve the X and Y chromosomes include Klinefelter's syndrome (47, XXY) and Turner's syndrome (45, XO). Chromosomal nondysjunction results in a number of trisomy syndromes that also have genitourinary implications (Table 1–6).

Developmental Abnormalities

There are also developmental abnormalities that may or may not have some underlying inherited basis that are often associated with genitourinary abnormalities (Table 1–7). The relationship of many of these problems, e.g., congenital, nonfamilial aniridia or hemihypertrophy, with Wilms' tumor are well documented though difficult to explain. Others, such as the high incidence of urinary abnormalities with abnormal sacral development and imperforate

Table 1–7. Developmental Anomalies Associated With a High Incidence of Genitourinary Abnormalities (Radiographic Evaluation Recommended)

Hemihypertrophy	Wilms' and adrenal tumors
Congenital, nonfamilial aniridia	Wilms' and adrenal tumors
Vaginal agenesis	Pelvic kidney, solitary kidney
Congenital scoliosis	Renal ectopia
Sacral agenesis	Neurogenic bladder
Imperforate anus	Unilateral renal agenesis, renal ectopia
Tracheoesophageal fistula	Imperforate anus (see above)
Myelomeningocele	Neurogenic bladder
Malformed ears	Renal agenesis
Potter facies	Oligohydramnios, renal agenesis
Prune belly syndrome	Dilatation of renal collecting system, megalourethra, undescended testes