Urologic Problems in Anorectal Malformations

23

23.1 Introduction

Urogenital problems in patients with anorectal malformation represent a very important source of morbidity; in fact, it is more likely for a child with an anorectal malformation to die from a urologic problem, rather than from gastrointestinal problems or any other associated defect. Pediatric surgeons must keep in mind that the patient with an anorectal malformation represents a potential life-threatening problem originated in the urogenital tract, mainly kidney damage and eventually kidney failure.

The association of urogenital defects with anorectal malformations has been recognized for a long time. The incidence has been estimated to vary from 25 to 85 % [1–17]. The majority of reports inform of an incidence around 30–50 %. We believe that the difference found between different authors is a consequence of the thoroughness of the search. A high index of suspicion, in an institution with high technologic support, mostly likely will find more urologic problems.

Table 23.1 shows the frequency of associated urologic defects for each type of anorectal malformation. The anorectal defects are listed according to the degree of complexity, being the simplest the perineal fistulas and in the opposite extreme the cloacal exstrophy and other complex malformations. It can be very easily seen that the frequency of association increases directly proportional to the height of the defect and the complexity of the malformation. This information is very valuable to increase the index of suspicion of the clinician and allow an early detection of the urologic problems, particularly those that may produce renal damage and eventually kidney failure.

It is a relatively common experience to see a patient that was born with an anorectal malformation that was repaired early in life, and the parents were not alerted about the fact that the patient had a urologic abnormality, and years later, the parents have the very unpleasant experience of learning that the child needs a kidney transplant. In some cases, the patient is born with the kidneys already severely damaged, and the role of the physician should be directed to avoid worsening of that problem. On the other hand, some patients are born with normal kidneys, but the lack of care of a problem such as vesicoureteral reflux and a poor functional bladder eventually may damage the kidneys, and the patient may require a kidney transplant or even die. This last situation is mostly preventable and should not occur. All this means that it is very important to suspect, diagnose, and treat adequately all those urologic problems associated with anorectal malformations. In addition, patients with anorectal malformations should be followed on a long-term basis, monitoring their kidney function

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Table 23.1 Frequency of association of urologic defects for each type of anorectal malformation

Females			
	Absent kidney (%)	Vesicoureteral reflux (%)	Hydronephrosis (%)
Cloaca CC >3 cm	26	40	45
Cloacal exstrophy	26	21	9
Cloaca CC <3 cm	17	21	22
Vestibular	10	13	6
Perineal	4	5	5

Males					
	Absent kidney (%)	Vesicoureteral reflux (%)	Hydronephrosis (%)	Hypospadias (%)	UDT (%)
Bladder neck fistula	31	29	24	15	18
Prostatic	16	6	6	6	7
Bulbar	11	13	9	10	5
No fistula	7	7	4	3	10
Perineal	2	3	6	1	2

and the anatomy of the urinary tract, because we know that many of them have anatomic or functional problems with a tendency to deteriorate.

23.2 Neonatal Approach

If a pediatric surgeon has no experience in the diagnosis and management of urologic problems, he should work together with a pediatric urologist in the initial evaluation of a patient with anorectal malformation.

All newborn babies with anorectal malformations should have a kidney ultrasound, within the first 24 h of life and before any surgical intervention. Female babies, particularly those with cloaca (single perineal orifice), must have, in addition, a pelvic ultrasound to rule out the presence of a hydrocolpos.

We believe that a male baby with a normal kidney ultrasound, that is passing urine satisfactorily, has a normal sacrum, and no evidence of tethered cord can be operated on without any further urologic tests. On the other hand, if the baby has hydronephrosis, he will require further urologic studies, including a voiding cystourethrogram. We do not believe that the voiding cystourethrogram should be done routinely, in all male babies with anorectal malformations, because in our experience, it is not a good study to diagnose the location of the recto-urinary fistula. In addition, the urethral catheterization in babies with rectourethral fistulas sometimes is difficult, because the patients frequently have kinks or narrowing of the urethra, particularly at the junction between the rectum and the urethra.

If the baby has tethered cord and/or a very abnormal sacrum (sacral ratio less than 0.4), he is at risk of suffering from deterioration of the bladder function, which eventually may affect the kidneys and therefore is considered a high-risk patient from the urologic point of view, even if the initial kidney ultrasound is normal.

The female baby with a cloaca represents a particularly difficult challenge (see Chap. 16). Thirty percent of patients born with a cloaca have a hydrocolpos that may produce an extrinsic compression of the ureterovesical junction, which results in megaureter and hydronephrosis. As we mentioned in the Chap. 16, before considering drainage of the urinary tract with a nephrostomy or ureterostomy, the surgeon must drain the hydrocolpos and reevaluate the megaureter and hydronephrosis. After the hydrocolpos has been drained, the megaureter will improve or disappear, as well as the hydronephrosis. If the hydronephrosis or the megaureter persists, the baby must be subjected to further evaluation to consider the possibility of making a ureterostomy, nephrostomy, or vesicostomy. We have seen

many babies with cloacas, hydronephrosis, and megaureter, subjected to unnecessary ureterostomies, nephrostomies, and/or vesicostomies, in a futile attempt to improve the hydronephrosis, without draining the hydrocolpos (see Chap. 16).

There is a small group of patients with a cloaca that are born with a very narrow (almost atresia) common channel which interferes with the emptying of the bladder. After draining the hydrocolpos, an ultrasound may show that the bladder is still full and does not empty well. An attempt to catheterize the bladder may be extremely difficult; in those cases it is justified to do a cystostomy or a vesicostomy.

Another possible indication for a vesicostomy is the case of a baby with demonstrated massive vesicoureteral reflux, megaureter, hydronephrosis, and important risk factors for poor bladder function, such as poor sacrum and tethered cord.

The newborn baby should not be taken to the operating room until the surgeon has ruled out and treated important associated urologic problems. After this, the baby is taken to the operating room to perform either a colostomy or a primary repair of the anorectal malformation.

23.3 The Importance of the Colostomy Type from the Urologic Point of View

The location and type of colostomy may have important urologic implications. It is important for the neonatologist as well as the urologist to remember this (see Chap. 5).

The colostomy that we recommend is one created at the end of the descending colon and proximal to the sigmoid loop. The purpose of that specific location is dual, first, to avoid prolapse of the proximal stoma, since the descending colon is fixed, and, second, to leave enough distal bowel (loop of the sigmoid) to allow a successful pull-through. In addition, we specifically recommend to make the distal stoma (mucous fistula) very narrow in order to avoid prolapse, but still, we believe that is important to keep it open to be able to irrigate the distal bowel and to perform radiologic studies, most specifically, a high-pressure distal colostogram. We strongly recommend separating both stomas in order to be able to put a colostomy bag, including only the proximal stoma and not the distal one. When both stomas are too close together, the mother is forced to use a single bag to cover both stomas, which may produce fecal contamination of the distal bowel and consequently of the urinary tract. Loop colostomies, therefore, from our point of view, are formally contraindicated in anorectal malformations, because they represent a source of fecal contamination of the urinary tract.

During the creation of the colostomy, we specifically recommend the surgeons to wash the bowel distal to the mucous fistula until it is completely collapsed and cleaned of meconium. If a baby receives a colostomy and after the procedure is not doing well and suffers from frequent urinary tract infections and from episodes of acidosis, it is extremely important for the physician to look at the type of colostomy that the baby has, looking for an explanation for that behavior. A loop colostomy producing frequent urinary tract infections would require an operation to separate the stomas. If the surgeon did not wash the distal bowel, sometimes the accumulation of meconium, plus mucus passing through the fistula, produces urinary tract infections. Finally, when the colostomy was opened in the transverse colon (which we consider contraindicated), the meconium may pass into the urinary tract, but also urine passes from the urinary tract into the colon where it is absorbed, producing hyperchloremic acidosis that may interfere with the growth and development of the newborn baby. When a pediatric urologist is called to see a baby with this kind of problem, he should look at the type of colostomy that the baby has.

23.4 Most Common Urologic Abnormalities in Male Patients with Anorectal Malformations

23.4.1 Absent Kidney

The most common congenital urologic defect associated to anorectal malformations is an absent, dysplastic, multicystic, or nonfunctional

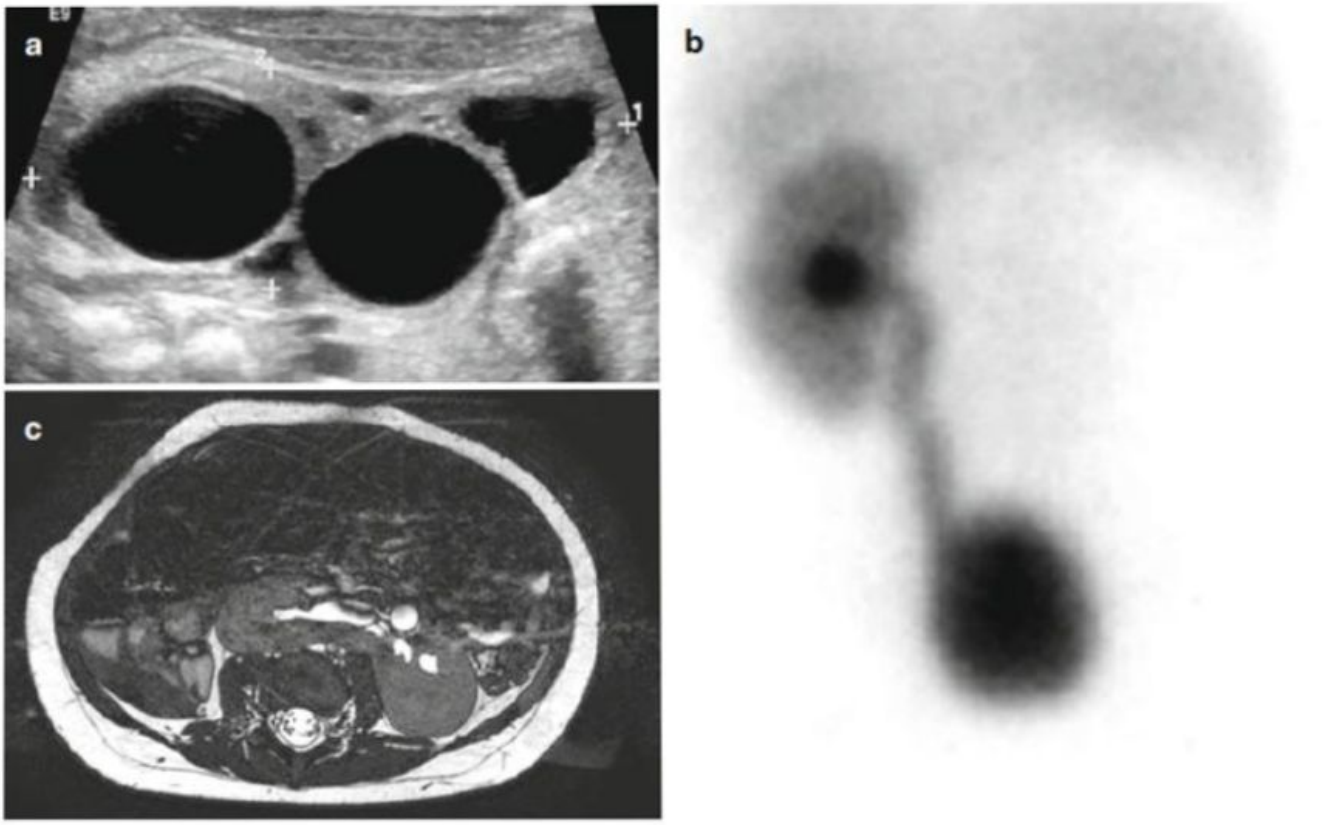

Fig. 23.1 Absent kidney (multicystic, dysplastic). (a) Ultrasound showing an image of a multicystic kidney. (b) Renal scan showing an absent kidney. (c) MRI showing a horseshoe kidney

kidney. In our series, 106 out of our 909 male patients and 165 out of 1,123 females were born with this problem.

Figure 23.1 shows the images of a patient with an absent kidney. Figure 23.1a shows the ultrasound and Fig. 23.1b shows a renal scan. This is by far the most common type of anatomic abnormality seen in anorectal malformations. The kidney may be totally absent or rather represented by a nonfunctional multicystic and/or dysplastic kidney remnant.

Figure 23.2a shows the images of an ultrasound in a patient with hydronephrosis, and Fig. 23.2b shows a case with hydronephrosis and megaureter in a patient with anorectal malformation. This problem was present in 198 out of 1,123 female patients and 70 out of 909 male patients. The presence of a megaureter in patients with anorectal malformations, most of the time, represents the existence of severe vesicoureteral reflux, since the cases of ureterovesical obstruction are very unusual. Of course, we must remember that cloaca patients may have an extrinsic compression of the ureterovesical junction that is released when a dilated vagina is drained.

Figure 23.3 shows the images of a voiding cystourethrogram of a baby suffering from vesicoureteral reflux (Fig. 23.3a unilateral and Fig. 23.3b bilateral). In our series, this problem was present in 207 out of 1,123 females and 133 out of 909 males. The incidence of reflux in cases of anorectal malformations has been estimated to vary from 30 to 60 % [18–21]. In our series it occurred between 14 and 18 % of the cases.

The presence of severe megaureter and vesicoureteral reflux in a newborn baby is an ominous sign. That baby will require special care to protect the kidneys, particularly in cases with real or potential neurogenic bladder, such as patients with severe sacral deformity and/or tethered cord. The best way to protect the kidneys in those babies is with a temporary vesicostomy, since trying to perform a ureteral reimplantation, with ureteral tapering, in an infant with a well-known poor functional bladder most likely will not succeed and may put the marginal renal function of the patient at risk.

There is one particular group of male patients born with a recto-bladder neck fistula.

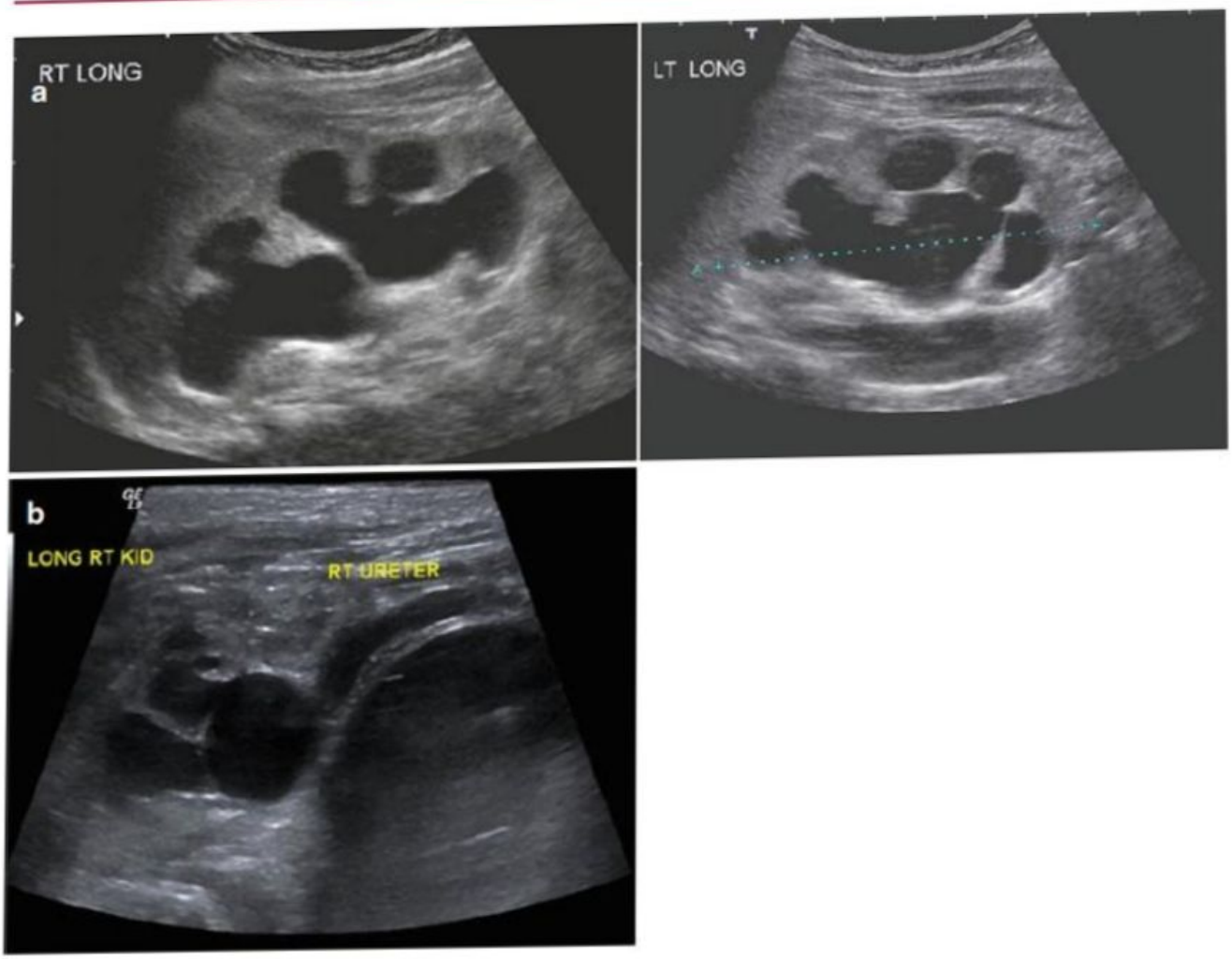

Fig. 23.2 Hydronephrosis (ultrasound image). (a) Bilateral hydronephrosis. (b) Hydronephrosis and megaureter

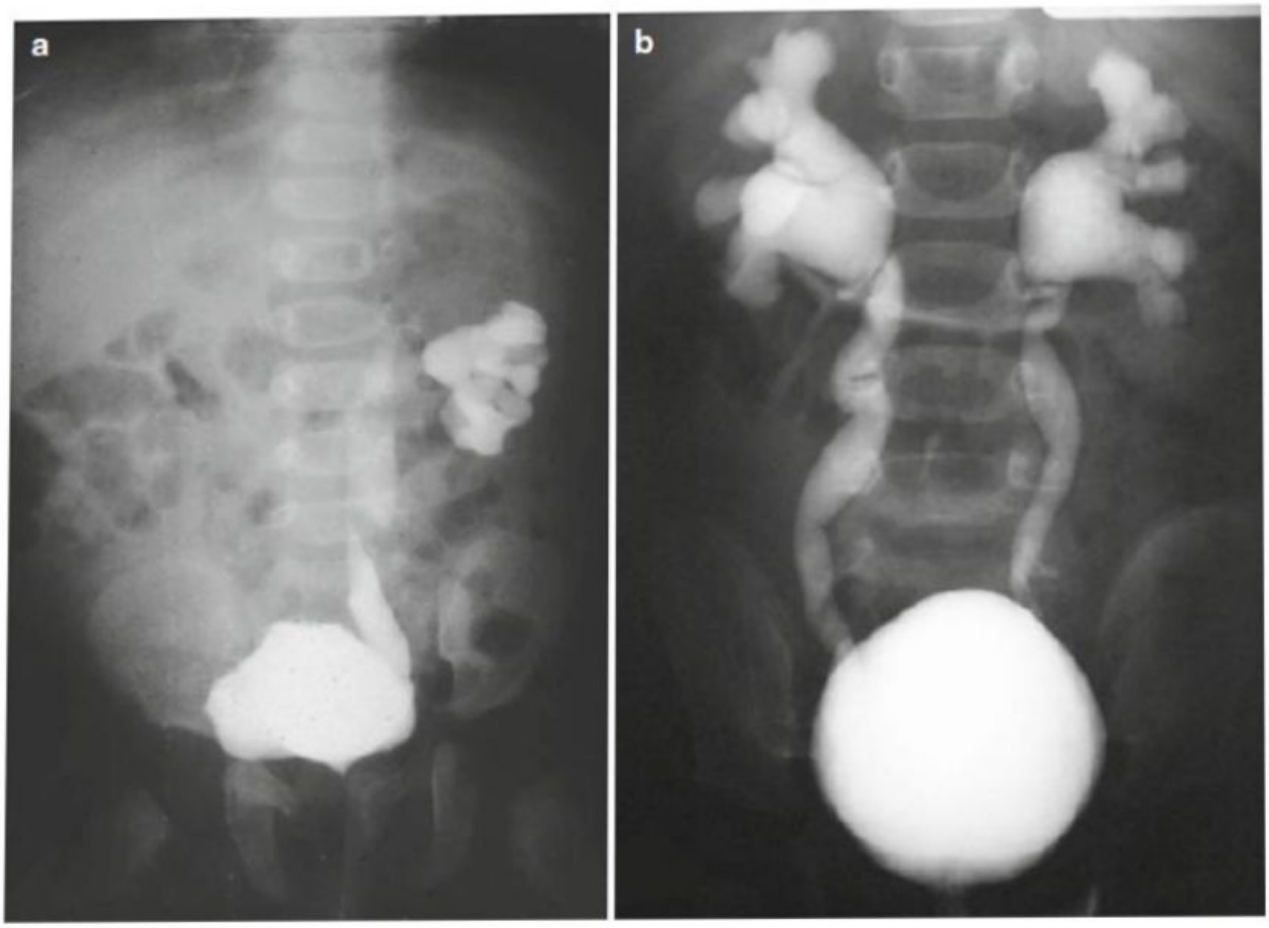

Fig. 23.3 Vesicoureteral reflux (VCUG). (a) Unilateral. (b) Bilateral

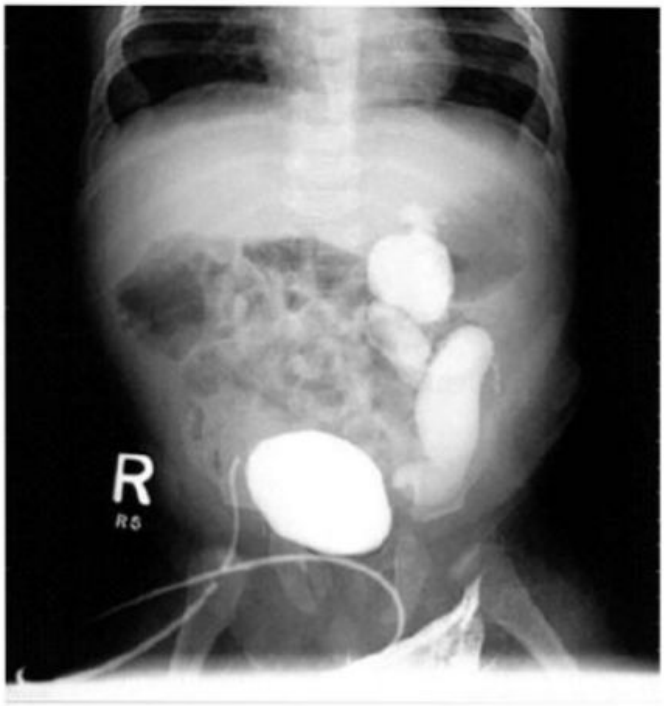

Fig. 23.4 VCUG in a patient with a single kidney with hydronephrosis and reflux

Twenty-five patients of a group of 111 patients born with a bladder neck fistula had an absent kidney, megaureter, and hydronephrosis in the opposite site (Fig. 23.4). These patients are at high risk to end up with a kidney transplant. The fact that a baby is born with hydronephrosis means that those kidneys had been suffering in utero and at birth already have a significant degree of damage. We have seen patients with hydronephrosis at birth and with a normal creatinine; they maintain the normal level of serum creatinine during the first years of life, but when they have a growth spurt, sometimes the creatinine suddenly elevates, indicating that most likely the patient was born with a limited kidney function, which became obvious when the patient grows significantly. Those cases with bladder neck fistula, single kidney, megaureter, and hydronephrosis should be considered high-risk patients.

The management of vesicoureteral reflux in patients with anorectal malformations deserves a special comment. The general principles of management of this disorder, in patients without anorectal malformations, should not be extrapolated and applied for cases with anorectal malformation for the following reasons:

A. Patients with anorectal malformations frequently suffer from a spectrum of sacral abnormalities that have a demonstrated effect on bladder function.
B. At least 25 % of patients with ARM suffer from tethered cord, which also has a significant negative effect on bladder function.
C. The repair of an anorectal malformation, when done inappropriately, produces different degrees of neurogenic bladder.
D. The repair of complex cloacas includes a significant dissection between the vagina(s), the bladder neck, the trigone, and the urethra. All these have a very significant effect on the bladder function. In addition, the scar produced by that dissection may represent a serious challenge to perform a reimplantation.

It is becoming obvious that the bladder function is a fundamental factor to take into consideration when making a decision about a ureteral reimplantation. A ureteral reimplantation, done in a patient with neurogenic bladder and incapacity to empty, most likely will fail. The decision to reimplant a ureter must not be taken without a previous urodynamic evaluation. This is particularly true in patients with megaureter and hydronephrosis. The ureteral reimplantation must be performed being sure that the patient is able to empty the bladder spontaneously or with intermittent catheterization through the urethra [22] or through an appendicovesicostomy tract (Mitrofanoff principle) [23, 24].

Because of all these reasons, in general, we try to avoid a reimplantation in a newborn with reflux, megaureter, and hydronephrosis. A vesicostomy is a better option, under those circumstances; it protects the upper tracts and gives us time to reevaluate the case and make an appropriate decision.

23.4.2 Urethral Problems

Figure 23.5 shows an image of male urethrogram of a patient born with an anorectal malformation and a recto-urinary fistula. The kink shown in the study most likely corresponds to the location of the fistula. However, we believe that the VCUG is not the ideal study to determine the location of the fistula. Some patients suffer from a urethral stricture

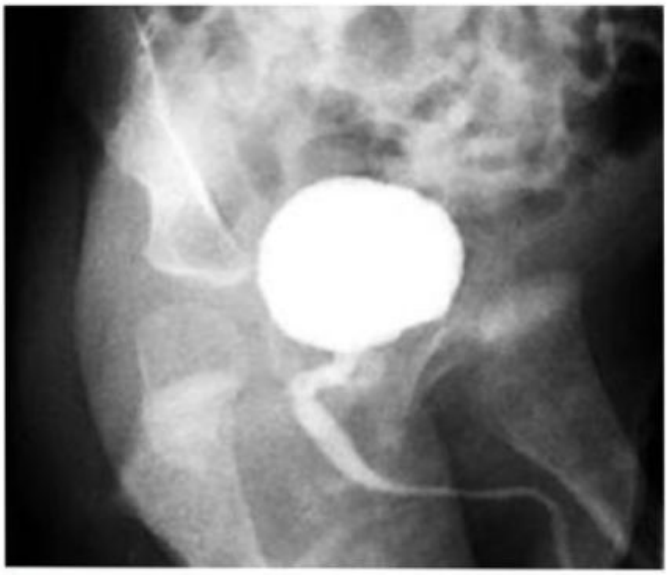

Fig. 23.5 Radiologic image of a kinked urethra. The narrowing and/or the kink is located at the junction of the rectum and the urethra (fistula site)

or a kink of the urethra at the same location of the junction of the rectum (fistula) (Fig. 23.6). The posterior sagittal approach represents an ideal way to repair urethral strictures. This can be done at the same time of the repair of the anorectal malformation. We have operated on our patients with this kind of problem and found relatively simple the urethral repair. The results have been good. Figure 23.7 shows operative pictures of a repair of a urethral stricture located at the same location of the fistula. A urethroplasty was performed using a flap of rectal tissue (Figs. 23.7a–d).

It is relatively common to see cases that have a very acute kink of the urethra at the same location where the rectum joins the urinary tract. When the doctors, radiologists, or surgeons try to pass a catheter, the catheter stops at that place or rather goes into the rectum. To try to overcome this obstacle, it is recommended to use a coude type of catheter (curved tip) to get access to the bladder. Sometimes, however, even with the use of this catheter, it is not possible to catheterize the bladder. In such cases it is necessary to pass the cystoscope using a wire in order to catheterize the bladder. There is a maneuver that proved to be useful to pass a Foley catheter under those circumstances. A lacrimal probe is inserted in the hole of the catheter, and the lacrimal probe is curved at its tip and can be externally directed (see Chap. 9). If the patient, in addition, suffers

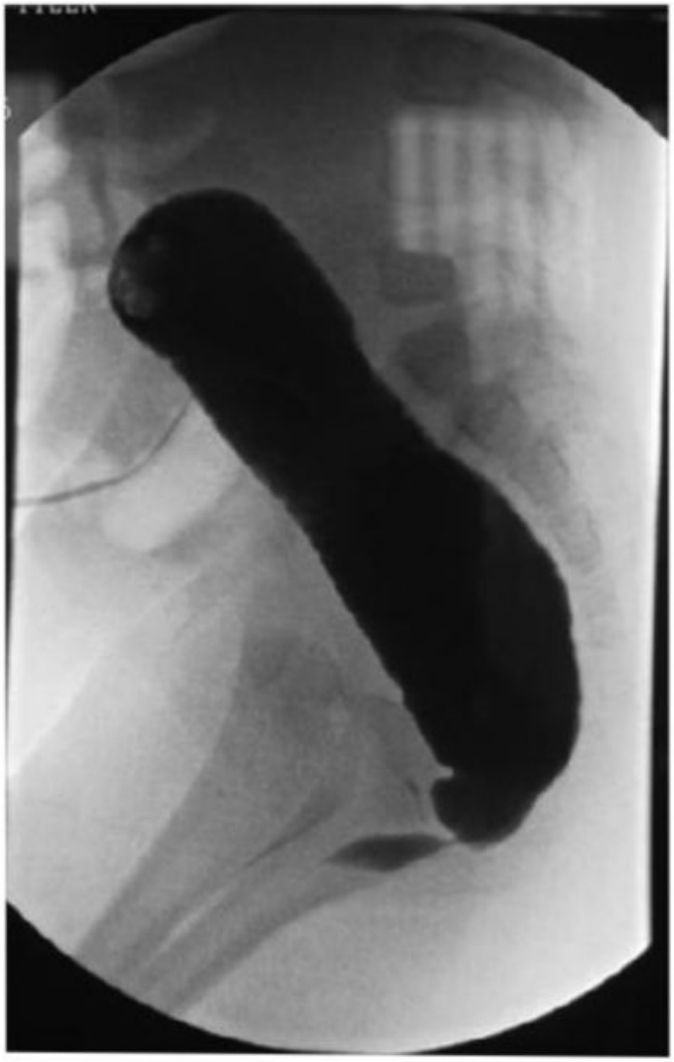

Fig. 23.6 Distal colostogram showing a urethral stricture at the junction of the rectum with the urethra

from a urethral stricture, then there is no way to pass the catheter into the bladder. Under those circumstances we allow the catheter to go into the rectum and approach the patient posterior sagittally; when the rectum is opened, one can see the catheter inside, and then under direct vision, one can redirect the catheter into the bladder. If the surgeon finds that there is a urethral narrowing, we have been using a little piece of the local rectal tissue to create a small flap to enlarge the area of the stricture of the urethra with good results (Fig. 23.7).

23.5 Bifid Scrotum

In our series of 909 cases of male patients with anorectal malformation, we found 102 patients suffering from a bifid scrotum (Fig. 23.8). Most

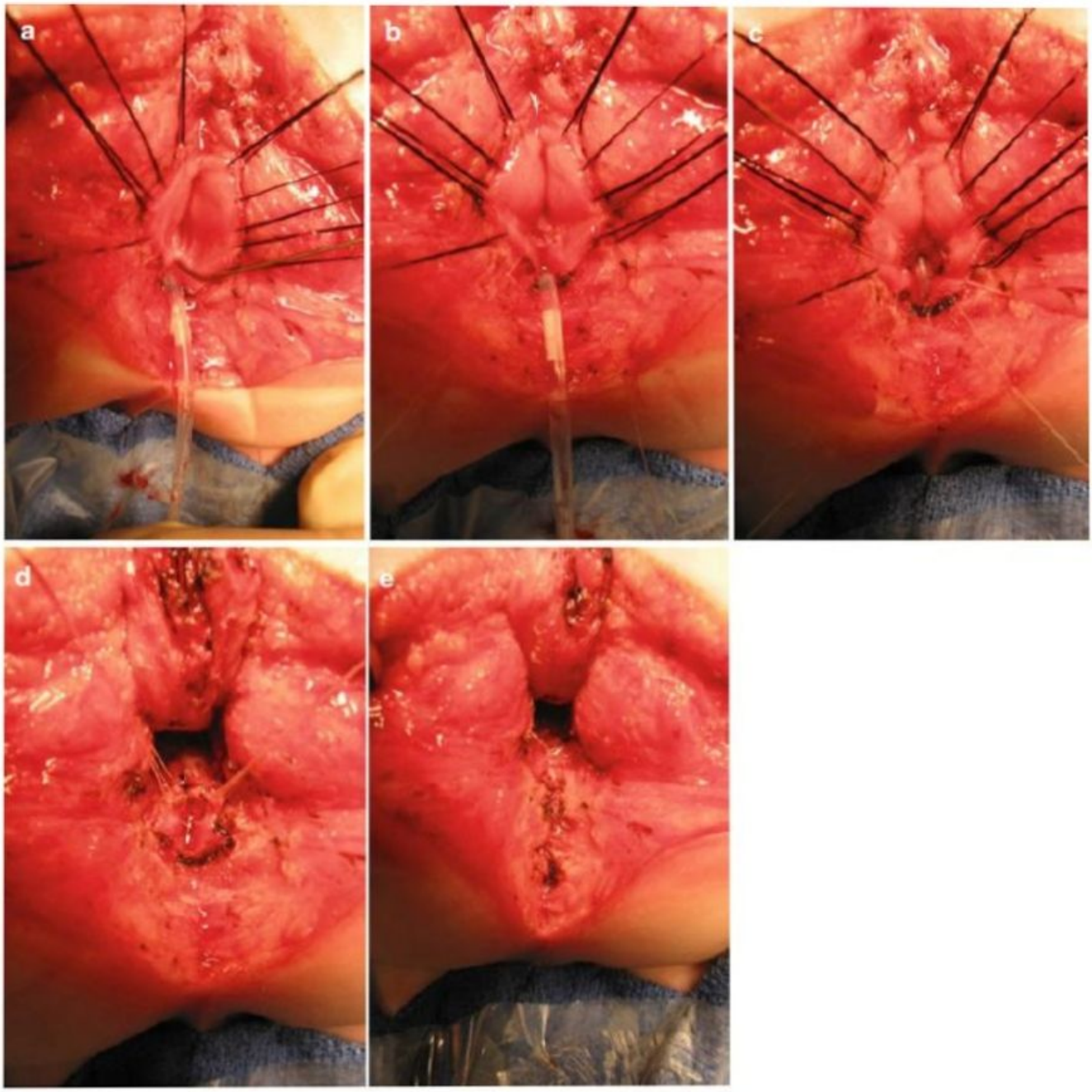

Fig. 23.7 Operative findings in a patient with urethral stricture. The catheter was redirected into the bladder under direct vision, and the urethral stricture was repaired. (**a**) The rectum open, lacrimal probe into upper the urethra, catheter emerging from the bulbar urethra. (**b**) The upper portion of the urethra has been enlarged with a plasty. (**c**) The urethral catheter was passed toward the bladder. (**d**) The area of stricture is expanded using rectal tissue. (**e**) Reconstruction finished

of the cases of bifid scrotum occur in babies who have anorectal malformations that belong to the "bad" side of the spectrum, in other words, patients that have bladder neck or high prostatic fistulas. Therefore, a bifid scrotum should be considered as a sign of a potential complex malformation.

Through the years, we learned to repair the bifid scrotum in a rather simple way, at the same time that we repair the anorectal malformation. It is important to recognize that in cases of bifid scrotum, there is a midline portion of nonelastic skin that separates both hemiscrotums (Fig. 23.9a–e). The skin of the scrotum is recognized because it has rugae and is elastic. The skin in between the hemiscrotum lacks that kind of rugae, is rather smooth, and is nonelastic. The technique that we use consists of resecting

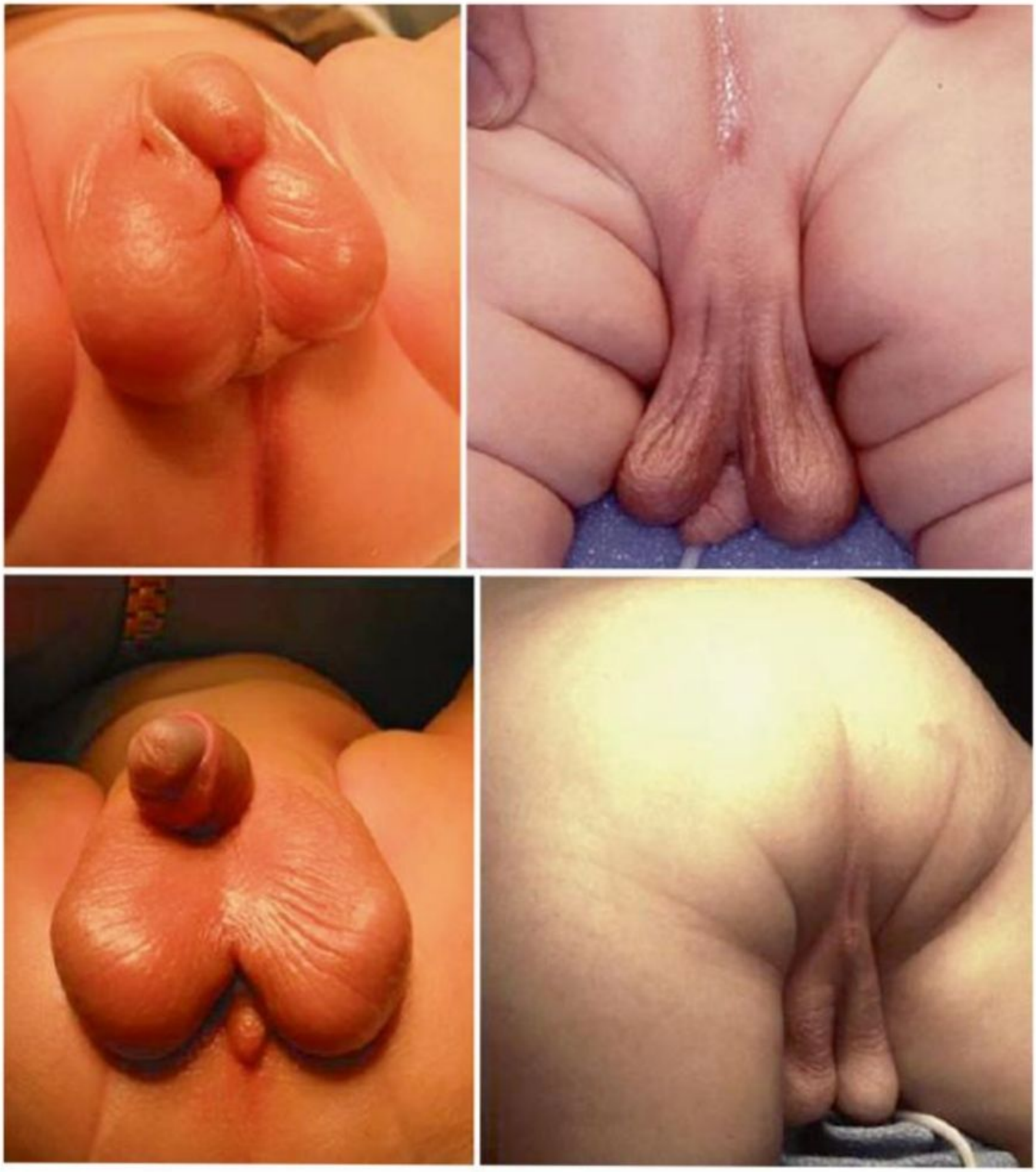

Fig. 23.8 Different types of bifid scrotum

the smooth, nonelastic portion of the skin. Figure 23.9a shows how we have marked the limits that separate the smooth, nonelastic skin from the normal rugae scrotal skin. Figure 23.9b shows the incision that is made with the needle-tip cautery. Figure 23.9c shows that the smooth skin has been resected and the beginning of a two layer suture of the skin edges (Fig. 23.9d). Both layers sutured with multiple, interrupted, 6-0 long-term absorbable sutures. Figure 23.9e shows the final aspect of the repair. Later in life, it is very difficult to know that this patient had a repair of a bifid scrotum and the parents have expressed their satisfaction.

chordee. In order to do that, the surgeons have to work on the common wall between the rectum and the urethra and move, if possible, the urethra a little forward. The idea is to create a reasonable space between the anal opening and the urethral opening and move the penis forward, away from the anal opening. After this repair, the urologist will take care of the problem of repairing the rest of the perineal urethra, as well as the penile urethra. Figure 23.10d shows the external aspect of a patient that underwent a repair of an anorectal malformation and separation of the rectum from the urethra in a severe perineal hypospadias.

23.7 Ectopic Ureters in Males

This malformation has been reported in cases without ARM [25]. It seems to be relatively rare. We believe that it occurs more often in patients with anorectal malformations.

Twenty-one of our male patients suffer from some form of ureteral ectopia. In patients with anorectal malformations, we have seen that the ureters have a tendency to open ectopically, in the line that goes from the normal location of the ureter in the trigone toward the bladder neck. It is relatively common to see patients with a ureteral orifice located closer to the bladder neck. There are however cases in whom, definitely, we see the ureter opening in the bladder neck (Fig. 23.11a). The opening of the ureter in the bladder neck frequently is associated with some degree of ureteral obstruction and megaureter. In addition, the presence of the ureter in that abnormal location may interfere with the closure of the bladder neck, producing a certain degree of bladder neck incapacity to hold the urine. We have relocated the ureters out of the bladder neck with success, recovering urinary control in patients that were leaking urine.

The most severe type of ureteral ectopia in males occurs when the ureters are connected to the posterior urethra (Fig. 23.11b). In these cases, the patient suffers from severe megaureter, and they frequently have severe damage to the kidney that usually ends up with a nephrectomy. The surgeon should be alerted about the fact that the megaureter connected to the posterior urethra may give the false intraoperative impression of the rectum. In one specific case, the patient was operated in another city without a distal colostogram, and the surgeon entered posterior sagittally looking for the rectum, found a "tubular structure," separated it from the posterior urethra, and pulled it down, believing it was the rectum, and it turned out to be a megaureter. The patient had actually a recto-bladder neck fistula.

As expected, the frequency of ectopic ureter is higher in patients with higher and more complex malformations. If one already made the diagnosis of an ectopic ureter located in the posterior urethra, the posterior sagittal approach represents a golden opportunity to identify the megaureter, separate it from the posterior urethra, close the posterior urethra, and push the megaureter up into the pelvis, to be recovered through a small incision in the groin, where it can be connected to the skin as a ureterostomy. It will take several months for us to determine the degree of renal damage that the kidney has and whether or not the patient will benefit from a nephrectomy or a ureteral reimplantation.

23.8 Ectopic Ureters in Females

All that we said about the ectopic ureters in the trigone and bladder neck is true for female patients. The ureters also may be ectopically connected to the urethra in a female. Figure 23.12 shows a picture taken during a transpubic approach of a female patient with a complex malformation. It would be very difficult to separate the ureter with a different type of approach. We use the transpubic approach in cases that we believe would be very difficult to approach in a different way.

In patients with cloaca, it is also relatively common to find ureters connected ectopically to the vagina. Again, most of the time these ureters belong to severely damaged kidneys.

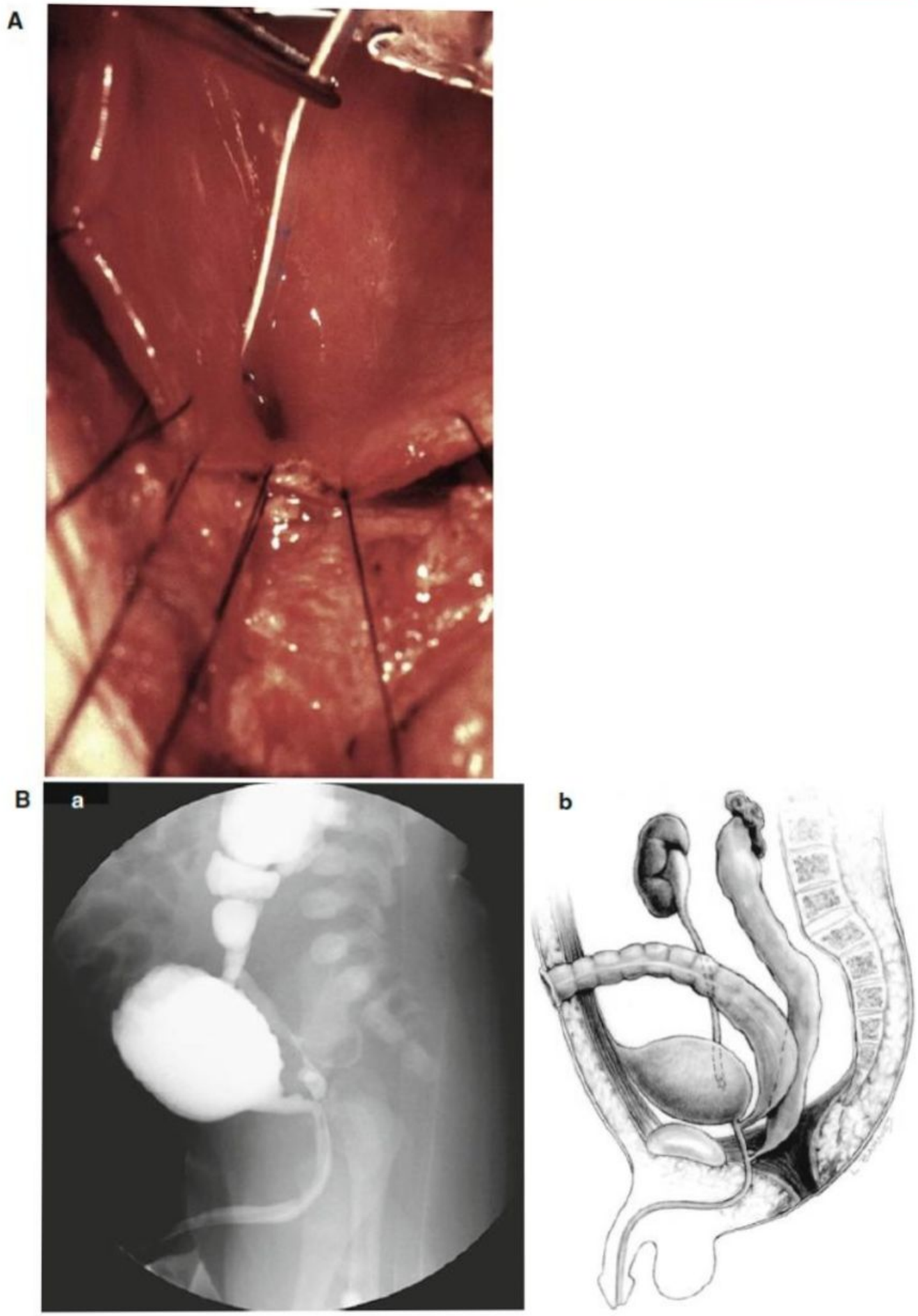

Fig. 23.11 Diagram showing ectopic ureters. (**A**) Connected to the bladder neck. (**B**) Connected to the posterior urethra. (a) Voiding cystourethrogram. (b) Diagram

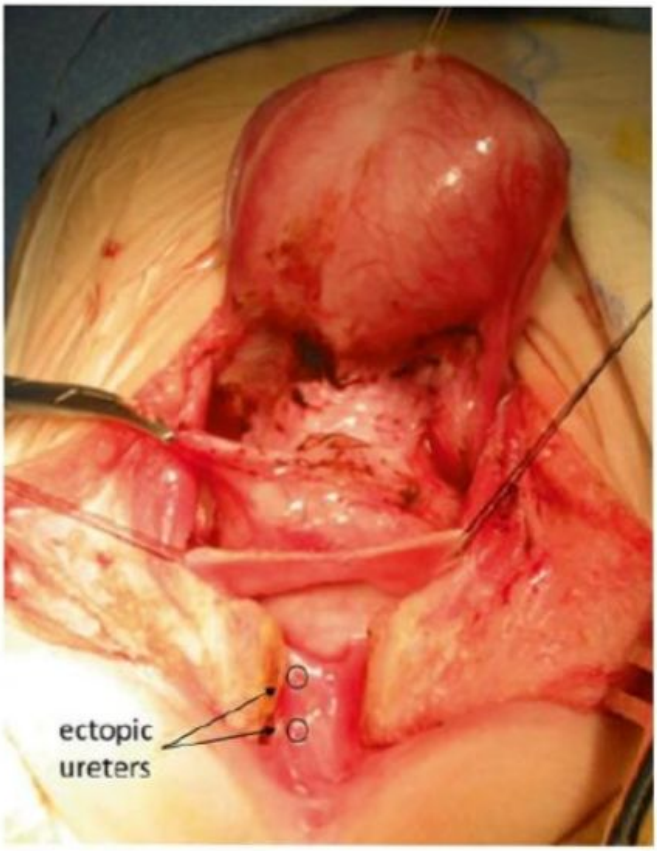

Fig. 23.12 Ectopic ureters in the urethra of a female patient with a complex malformation. Intraoperative picture taken during a transpubic approach

23.9 Ectopic Vas Deferens

This rare anomaly has been reported in patients without anorectal malformation [26–28]. The most common clinical manifestation has been orchiepididymitis. However, we found more publications related to this malformation in cases of anorectal malformations [29–38].

We have demonstrated the presence of ectopic vas deferens in three cases of our population of male patients with anorectal malformations. We suspect that this defect occurs more often, in patients with ARM, particularly in the group of patients who suffer from orchiepididymitis. It is not easy to demonstrate this problem. We were able to make the diagnosis injecting contrast material directly into the vas deferens with a 30-gauge needle. The vas deferens may be abnormally connected to the bladder or to the ureter. Figure 23.13a shows an operative picture of an ectopic vas deferens, connected to the ureter. The symptomatology in this patient consisted of recurrent orchiepididymitis. The treatment for

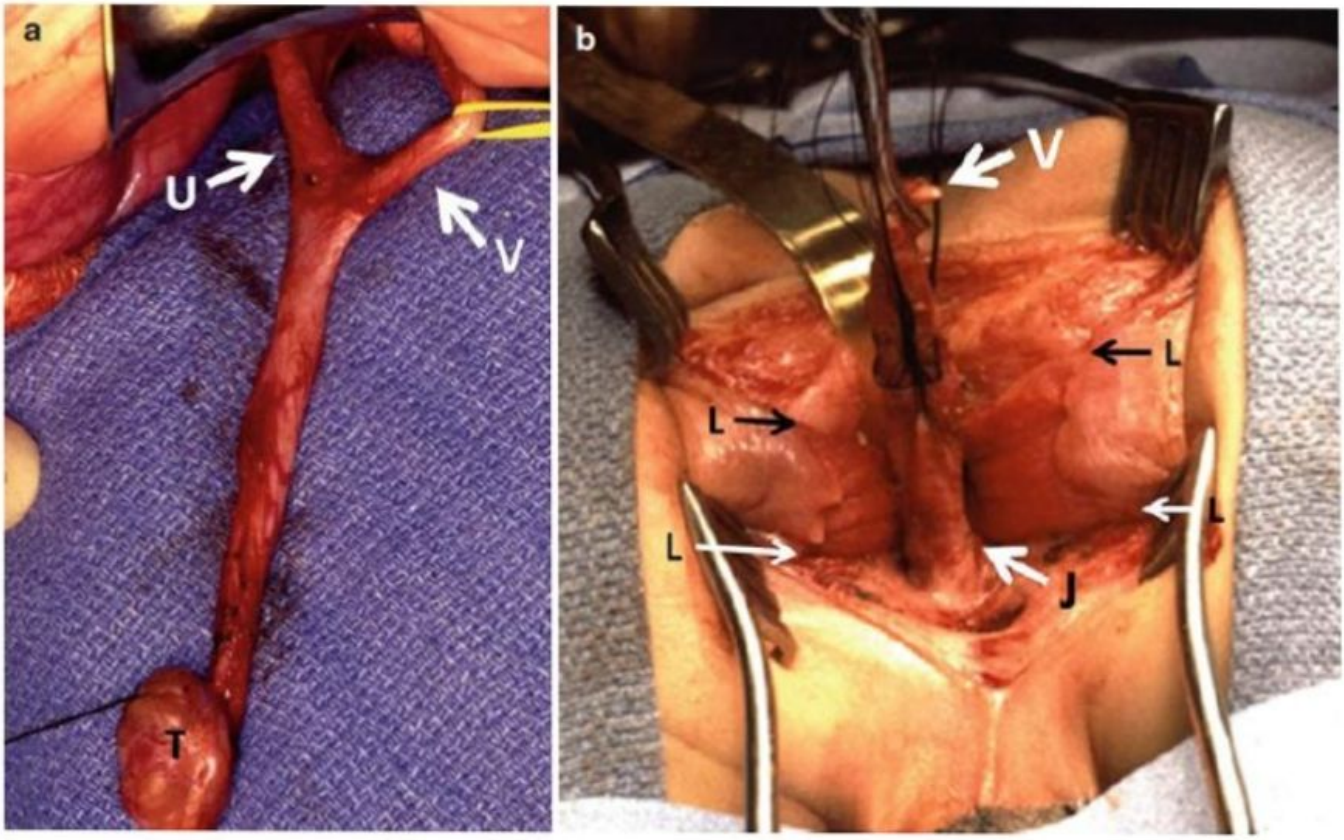

Fig. 23.13 Ectopic vas deferens. (**a**) Operative picture *V* vas deferens, *U* ureter, *T* testicle. (**b**) Vas deferens connected to a giant seminal vesicle – picture taken during the trans-anorectal resection of the giant seminal vesicle. *V* vas deferens, *J* junction of seminal vesicle to urethra, *L* limits of the anus

this condition should be a vasectomy, to preserve the endocrine function of the testicle that may be destroyed after repeated episodes of infection. Sometimes, the vas deferens is abnormally connected to an extremely dilated giant seminal vesicle which is also a source of orchiepididymitis (Fig. 23.13b).

Orchiepididymitis occurs relatively frequent in patients with anorectal malformations. Many patients who underwent a repair of an anorectal malformation at our center were followed by other colleagues in other cities. Some of those patients suffered from an episode of "acute scrotum" and were surgically explored to rule out a possible testicular torsion, only to find that the babies actually suffered from orchiepididymitis. In fact, we have never seen or heard of a case of testicular torsion in a patient with ARM. Yet, we have seen 17 cases of orchiepididymitis. The etiology of this condition seems to be multifactorial. Most patients have a urinary tract infection and some degree of neurogenic bladder. Some of them have an identifiable anatomic problem that may explain the problem. These include urethral obstructions and ectopic vas deferens. A patient with acute scrotum and ARM must be considered as orchiepididymitis until proven otherwise.

All patients with orchiepididymitis deserve a full urologic evaluation after the acute episode. In addition, they must receive prophylactic medication to try to avoid episodes of urinary tract infections. The anatomic problem must be treated when found. In cases of frequent recurrences, a vasectomy of that particular side is indicated, in order to try to preserve the endocrine function of that testicles; otherwise, it may be totally destroyed.

23.10 Ectopic Verumontanum

An unreported, unusual, interesting malformation is the ectopically located verumontanum. We have four cases suffering from this defect within the group of patients born with recto-bladder neck fistulas. We become aware of this condition because one of our first patients operated from a bladder neck fistula reached adolescence and came for consultation, because even when he had erection and orgasm, he did not ejaculate. Urinalysis after ejaculation demonstrated the presence of sperm in the urine. This was not what is called retrograde ejaculation, because by cystoscopy we documented the presence of the verumontanum in the trigone (Fig. 23.14). After that case, we intentionally started looking for the location of the verumontanum in patients with bladder neck fistula, and we were able to find three more cases. This number of cases only represents 30 % of the patients who underwent an endoscopy. Most of the patients operated on for bladder neck fistula have not reached adolescence yet, so we expect that in the coming years more of these patients will come looking for help under these circumstances.

We now follow the routine of performing cystoscopy in all male patients with anorectal malformation, during the same anesthesia that we used for repair of the malformation. This is particularly important in patients with a bladder neck type of malformation in which this defect occurs.

We have been told that these patients may be able to fertilize their couple by retrieving the sperm from their urine and artificially inseminate the future mother. None of our patients have done that so far.

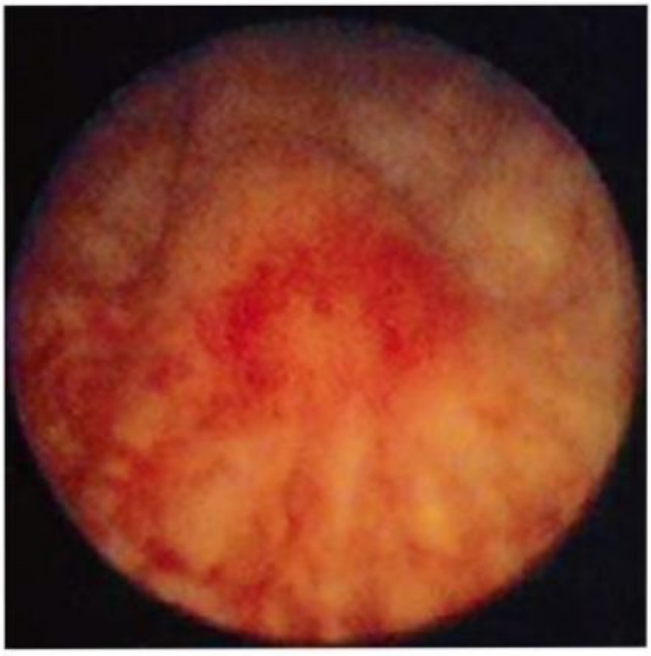

Fig. 23.14 Ectopic verumontanum – cystoscopic appearance

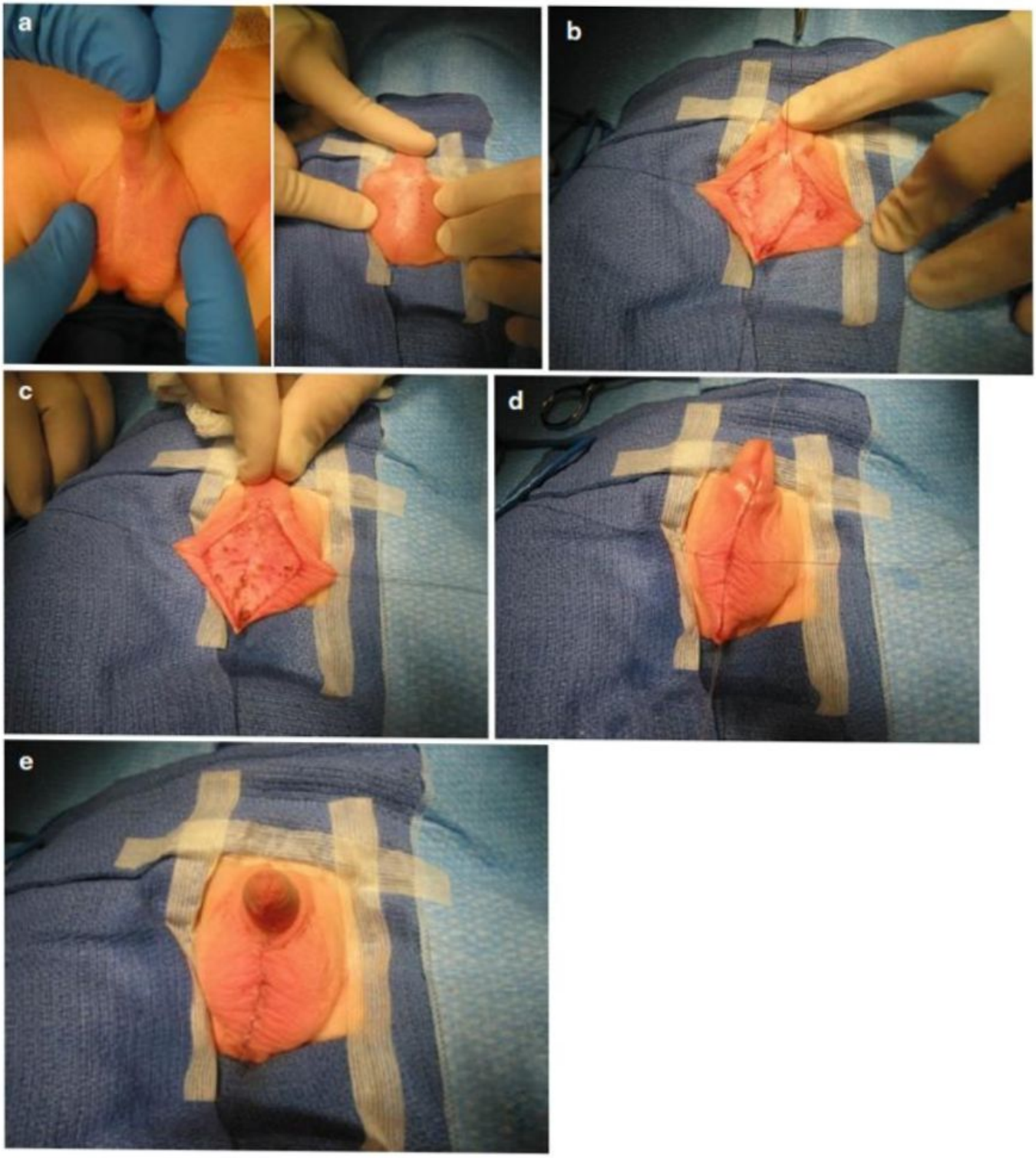

Fig. 23.9 Repair of a bifid scrotum. (**a**) Marking the limits of the abnormal skin. (**b**) Incision. (**c**) Resected non elastic skin. (**d**) Two-layer suture. (**e**) Final aspect

23.6 Hypospadias

Sixty of our male patients suffered from different types and severity of hypospadias. It may go from a subcoronal type (Fig. 23.10a) to an extreme type in which the urethral orifice is located immediately anterior to the anal opening (Fig. 23.10c). Extreme, severe types of hypospadias are frequently associated to penoscrotal inversion.

The timing for the repair of hypospadias is something rather controversial. Some pediatric urologists like to repair the hypospadias

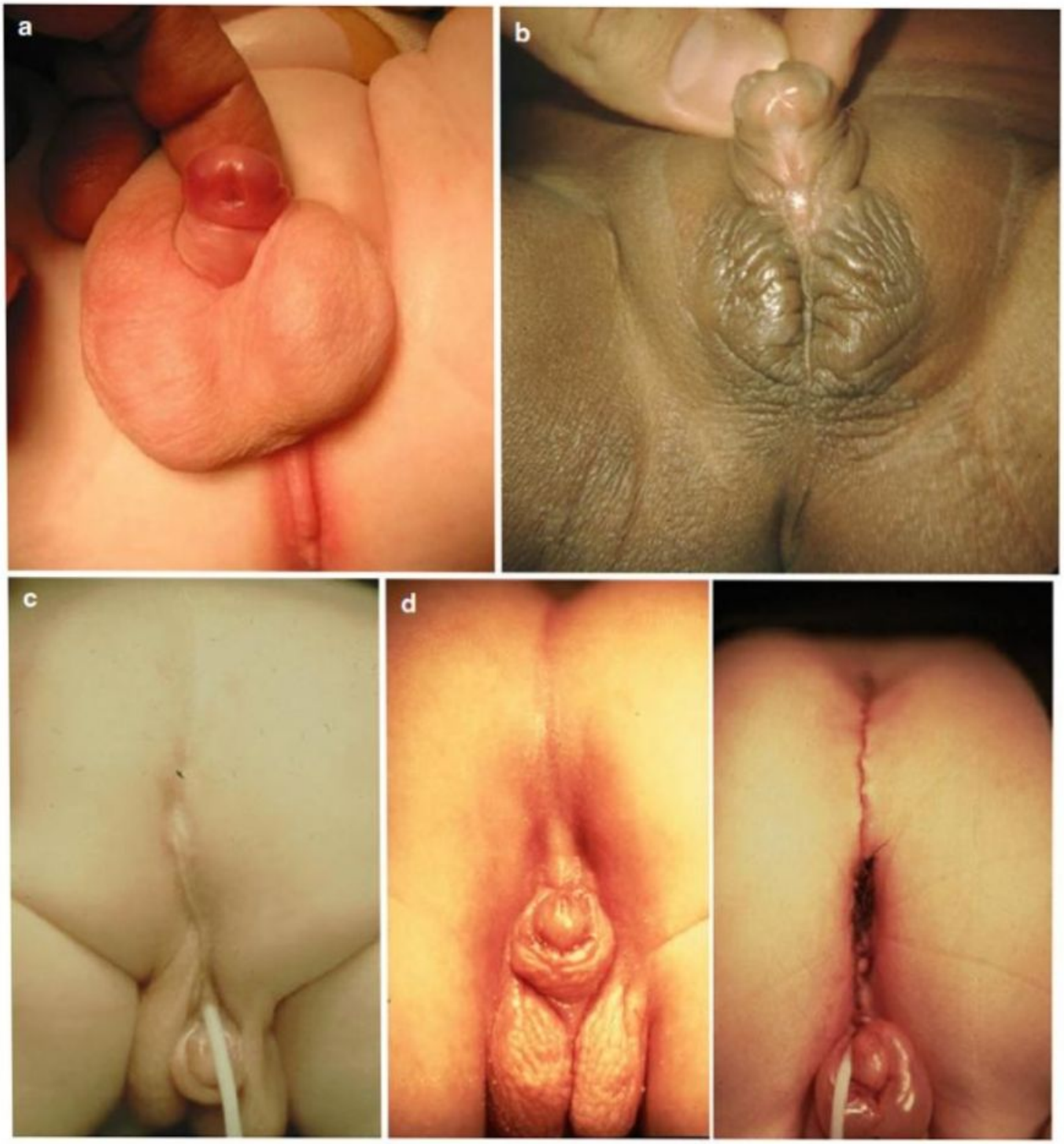

Fig. 23.10 Hypospadias. (a) Subcoronal. (b) Mid-shaft. (c) Perineal. (d) Repaired anorectal malformation with severe hypospadias. The urethra was originally located next to the anal opening. The urethral opening was moved forward, and the anus was moved back

before the colostomy is closed, to avoid fecal contamination during the postoperative period. Some others prefer to wait until the patient is older to be able to deal with better tissues and have more chances of success. From the point of view of the pediatric surgeon, it is important to identify the most severe type of hypospadias (Fig. 23.10c, d) in which the urethral orifice is located immediately anterior to the anal opening. The surgeon must keep in mind that the rectum and urethra share a common wall, usually in the area of the spongiosum tissue. Those babies have a strictured anal opening located anterior to the center of the sphincter, and therefore, the operation will consist in moving the anus back and simultaneously to correct the severe

23.11 Megalourethra

This malformation has been reported in cases without anorectal malformation [39, 40], as well as associated to anorectal malformations [41–47]. The patients that we have seen have a normal-looking penis (Fig. 23.15a); it is necessary to retract the foreskin to be able to see the abnormality (Fig. 23.15b). This means that this is the kind of defect that requires a good index of suspicion in order to make the diagnosis.

These patients are born with a very large urethral meatus, followed by a very dilated penile urethra. From the base of the penis to the glans, the urethra is extremely wide. However, from the base of the penis to the bladder, the urethra is otherwise normal. We have not seen many of the severe types of megalourethra with the absence of corpora and a large saccular floppy penis, described in the literature. Our cases were not associated with hypospadias. The repair of this defect is much easier than the one in hypospadias, the reason being that we are dealing with very good tissue and plenty of urothelium to repair the urethra. With the foreskin completely retracted back, like in a circumcision, we open the urethra ventrally (Fig. 23.15c) and excise the extra tissue of the megalourethra. The urethra is reconstructed over a convenient-sized Foley catheter (Fig. 23.15d). Figure 23.15e shows the final appearance of a patient with this defect after repair. We have 15 cases, all of them repaired with the same technique prior to the closure of the colostomy, with excellent results. These patients never developed any fistula, and they have normal voiding. Figure 23.15f, g shows a case of a much larger, floppy urethra.

23.12 Ureterovesical and Ureteropelvic Obstruction

We believe that most likely these two congenital abnormalities occur in cases of anorectal malformation with similar frequency as in the general population. It is extremely unusual to detect them in cases of anorectal malformations. The treatments of these defects are well described in the textbooks of urology.

23.13 Neurogenic Bladder

Neurogenic bladder associated to anorectal malformations is receiving significant attention [48–64]. The subject is not easy to study, considering the following facts:

A. May be congenital
B. May be acquired
C. May be influenced by the complexity of the anorectal malformation
D. Is influenced by the sacral defect
E. Is influenced by the tethered cord

Most of the publications include a rather small number of cases of different malformations with or without sacral defects or tethered cord. All this makes the analysis and interpretation of those publications a rather difficult task.

Most authors, like us, agree that it will be ideal to do a urodynamic evaluation pre- and postoperatively in all cases, documenting the sacral ratio and the presence or absence of tethered cord in order to draw valid conclusions.

We believe that most pediatric surgeons and pediatric urologists are now aware of the relevance of the prevention, early diagnosis, and management of neurogenic bladder in order to protect the kidneys.

Intermittent catheterization and bladder augmentation had a definitive positive impact to protect the kidneys in patients with hypertonic, neurogenic bladder [65–70]. However, these are patients who must be followed for life. The bladder augmentation has short- and long-term significant morbidity that must be detected and treated efficiently [71–78].

So far, in our series 58 patients underwent a bladder augmentation, 32 of them are performed by the senior author until 2005, and 26 since that time are performed by pediatric urologists. Most of these operations were done in girls with cloacas.

Some patients with anorectal malformations have manifestations of a neurogenic bladder since birth (primary). That is understandable when the patients have a poor sacrum (sacral ratio less than 0.4) and the presence of tethered cord. However, a

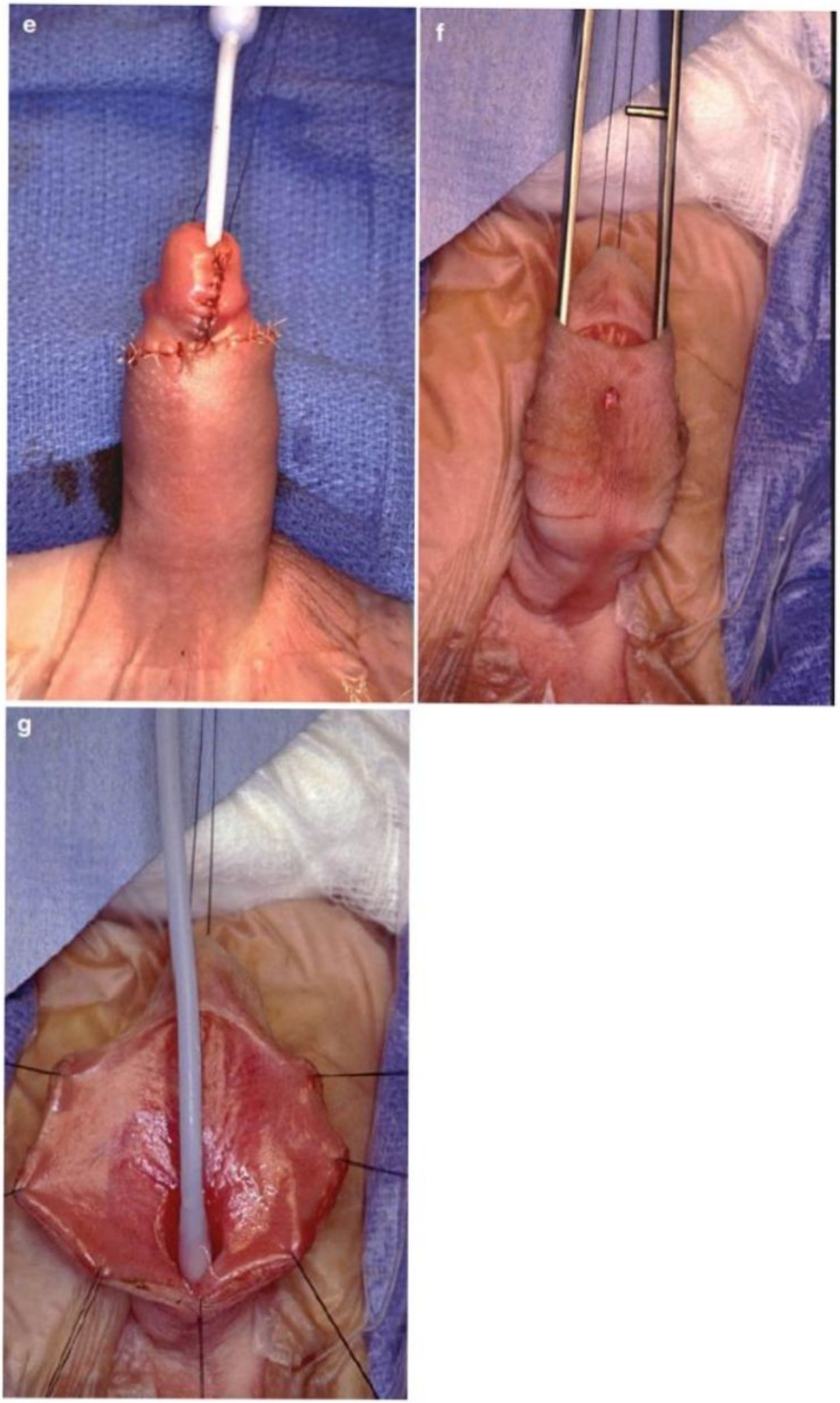

Fig. 23.15 (continued)

sagittal incision, in a technically deficient manner, frequently produces nerve damage that results in impotence and neurogenic bladder that was not present prior to the operation. The most important rule to follow during the posterior sagittal approach is for the surgeon to remain exactly in the midline. Once the rectum is found, the dissection of the rectum must be performed, remaining as close as possible to the rectal wall. Deviating from these rules increases the chances of nerve damage.

23.15 Posterior Urethral Diverticulum (Animation 13.2)

In our series of male patients, 32 of them had the anorectal repair done at another institution and came to us complaining of "urinary incontinence, urinary tract infections, and sometimes orchiepididymitis," and when we studied them, we found that they had a piece of rectum left attached to the urethra during the repair of the anorectal malformation. We called this "posterior urethral diverticulum" (Fig. 23.16). The retrospective analysis of these cases showed that most of these patients were born with a rectourethral bulbar fistula, and the surgeons tried to repair the malformation approaching it through the abdomen, following the old tradition of performing an abdominoperineal pull-through for those "high" imperforate anuses (we do not use that terminology anymore). While trying to dissect the rectum through the abdomen, trying to reach the bulbar urethra was very difficult, and frequently the surgeons decided to "play it safe" and amputated the rectum at a "convenient" level, leaving a piece of rectum attached to the urethra.

More recently, we have seen more cases of posterior urethral diverticulum in patients operated laparoscopically [82–87]. The surgeons, again, were trying to reach the lowest part of the rectum and its attachment to the bulbar urethra; they found that technically difficult and amputated the rectum again, leaving a piece of rectum attached to the urethra (Animation 23.1). Characteristically, these patients complained of passing mucus through the urethra in between voiding episodes. In addition, as can be seen in Animation 23.1, during the process

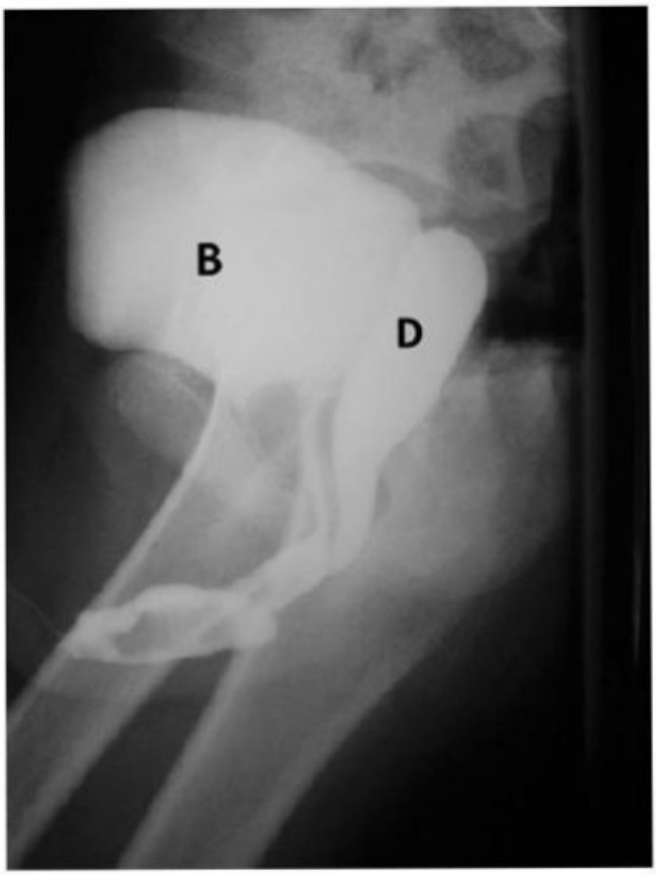

Fig. 23.16 Voiding cystourethrogram in a case of posterior urethral diverticulum, *B* Bladder, *D* Diverticulum

of voiding and emptying the bladder, part of the urine goes out through the penis, but other significant volume may go into the diverticulum. The patient finishes emptying his bladder and goes playing, and during that time, he leaks urine from the diverticulum.

The diagnosis of this condition requires a good index of suspicion. The history of being born with a rectourethral bulbar fistula operated through the abdomen (open or laparoscopically) is highly suggestive. In addition, the passing of mucus through the urethra and leaking urine in between episodes of voluntary voiding strongly suggest the presence of a posterior urethral diverticulum. The diagnosis can be confirmed with a voiding cystourethrogram (Fig. 13.1b) which may or may not detect the diverticulum. An MRI is a better study to see the diverticulum in a very accurate way (Fig. 23.16). Finally, a cystoscopy will confirm the diagnosis (see Chaps. 9, 13 and 22).

The treatment is straightforward. The diverticulum can be resected through a posterior sagittal approach. The pull-through rectum is

mobilized; the diverticulum is open and is separated from the urethra following the same steps and principles described for the separation of the rectum and the urethra in cases of rectourethral bulbar fistula.

One particular patient, 30 years old, developed an adenocarcinoma at the junction between the diverticulum and the urethra. Fortunately, we detected the case early enough to resect it and cure the patient. This is one of the reasons why we believe that the laparoscopic approach of a rectourethral bulbar fistula must be considered contraindicated.

23.16 Sexual Problems

Most of the male patients born with anorectal malformations operated by us that reached adulthood are sexually active. They claim to have satisfactory intercourse and many of them have children. There is however a small group of patients that suffer from impotence and others from lack of ejaculation [88, 89]. It is not clear to us why that happens. The lack of ejaculation may happen in patients that underwent "difficult" operations, performed by surgeons who were lost in the operative field, and we suspect that most likely a "small piece of tissue" that represents the prostate in babies was inadvertently resected. The majority of these adult patients complaining of sexual problems were operated with "blind techniques." In other words, the surgeons had a poor visualization or exposure of the junction of the rectum to the urethra. We believe that one of the great advantages of the posterior sagittal approach is the exposure of the intrinsic anatomy of these malformations, which allows us to avoid this kind of complications.

23.17 Tethered Cord

Approximately, 24 % of all of our anorectal malformation patients have tethered cord [90, 91]. Others have found a higher prevalence (34 %) [92]. The presence or absence of tethered cord seems to be important to determine the future prognosis in terms of urinary function [93]. The effect of tethered cord on bowel control has not been scientifically studied. The subject is still very controversial. In addition, there is no unified criterion related to the indications to release a tethered cord. There is also lack of agreement related with the precise diagnosis of tethered cord. Many of our patients were seen by different neurosurgeons, and some of them felt that the patient had tethered cord, and others believed that they did not have tethered cord. In addition, some of our patients had an ultrasound or an MRI study that showed no evidence of tethered cord, and yet they were operated as if they had tethered cord, because the radiologist and/or the neurosurgeons believed that the conus was "abnormally thick," the patient had a "lipoma in the tip of the conus," or "the conus was not mobile." Other anatomic variations have been described to make the subject more confusing, including tethered cord with transitional lipoma and "short spinal cord" [94]. Some neurosurgeons believe that detethering the cords is important to prevent problems with the motion of the lower extremities. This is not supported by others [95].

On the other hand, we have seen adult patients with tethered cord, and they play soccer and have urinary and bowel control! We have seen others, whose mothers indicated that the child was having some problems in walking and running, were operated for tethered cord, and the patients experienced an improvement in the function and motion of the lower extremities. Unfortunately, we have also seen patients that have had release of the tethered cord and suffered urinary retention immediately after the operation and subsequently developed severe signs of neurogenic bladder that were not present preoperatively. A couple of our patients become paraplegic after an operation for tethered cord!

Most radiologists consider that the presence of the conus medullaris below L2 plus a nonmobile conus means that the patient suffers from tethered cord (Fig. 23.17a, b). At present time, we recommend to do a spinal ultrasound during the first few days of life, in order to rule out the presence of tethered cord. If the study shows no

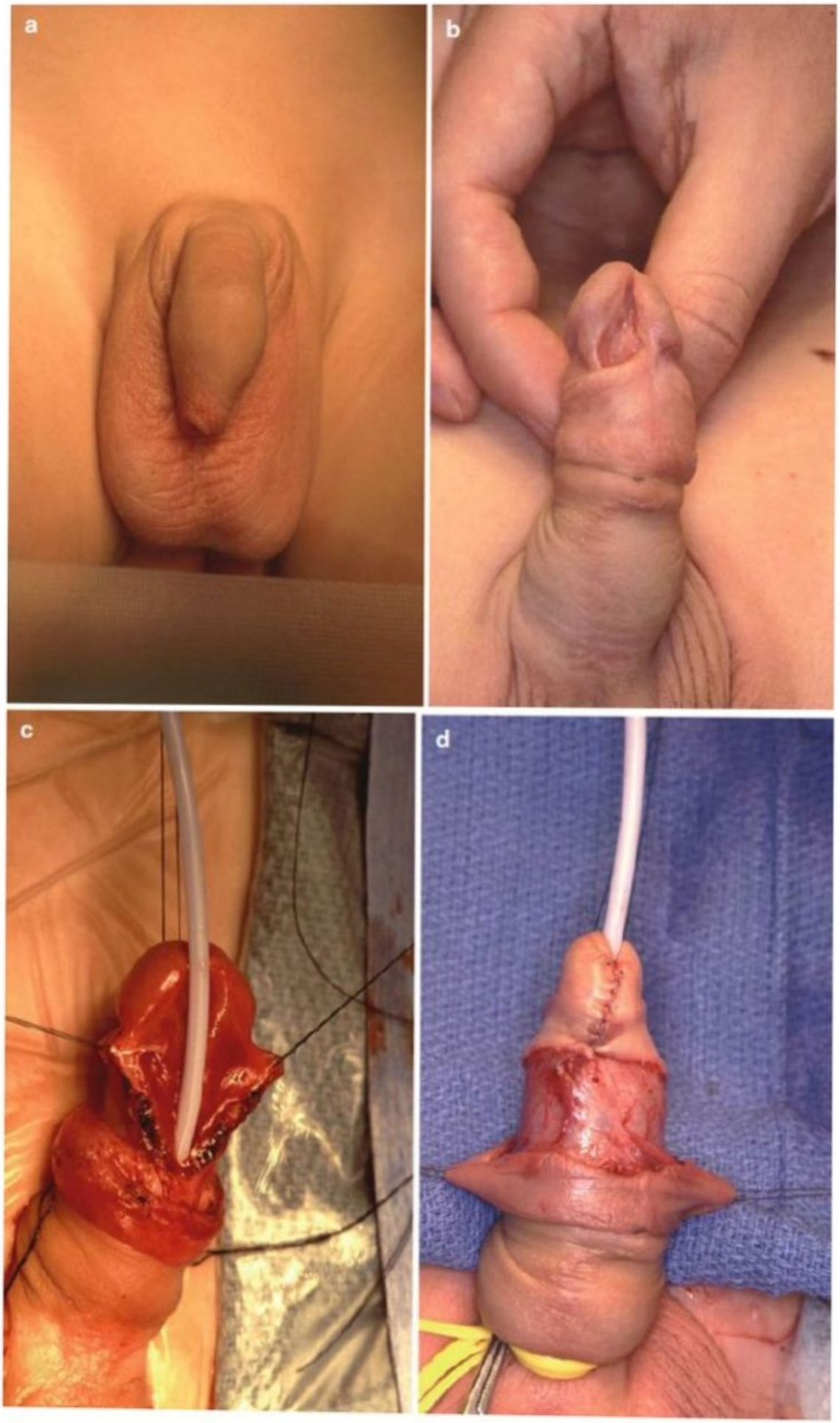

Fig. 23.15 Megalourethra. (**a**) External appearance – observe that it looks remarkably normal. (**b**) External appearance with the foreskin retracted showing the giant meatus. (**c**) The foreskin divided and retracted. The ventral portion of the megalourethra is divided. (**d**) The excessive urethral tissue is resected, and the urethra is reconstructed around a Foley catheter. (**e**) Final appearance. (**f**) Giant, floppy megalourethra. (**g**) Open giant megalourethra

large group of patients are born without neurogenic bladder, but they are subjected to a technically deficient anorectal operation, during which denervation is produced, and they acquire this problem.

In some cases, the nerve damage is immediately evident after the repair of the anorectal malformation. It is not uncommon to hear from the mother of the patient to say that the baby had erections prior to the operation and since the time of the operation she has not seen one! In other cases, the baby was passing urine satisfactorily, but suffered from urinary retention after the procedure. The urinary retention may be temporary; the Foley catheter is reinserted and removed after a week or two, and then the patient may start passing urine. Other times the damage is permanent. However, the fact that the baby passes urine does not mean that we can rule out the presence of a neurogenic bladder.

The most common bladder disorder in babies operated for anorectal malformations is the incapacity to empty the bladder. As part of the follow-up of all patients with anorectal malformations, we always order a kidney ultrasound and bladder ultrasound (taken at different postoperative intervals, for instance, 1, 3, 6, and 12 months). In patients in which we see a persistently full bladder, one must suspect that the baby is not emptying the bladder well. A high residual volume of urine reflects an incapacity to empty the bladder, all of which may eventually produce vesicoureteral reflux and kidney damage. We have also seen patients that do not have vesicoureteral reflux, yet a persistently full bladder may also produce megaureter and kidney damage. By emptying the bladder with a catheter and repeating the ultrasound, one can see the improvement of the megaureter and hydronephrosis which in the absence of reflux means that the megaureter and hydronephrosis are not provoked by a stenosis, but rather by extrinsic ureteral compression provoked by a distended bladder. These patients require a full urinary evaluation, including a urodynamic study and follow-up by a competent pediatric urologist who must decide whether the baby is a candidate for intermittent catheterization to help him empty the bladder. When the intermittent catheterization is not feasible or interferes with a good quality of life, some urologists perform a Mitrofanoff type of operation [23, 24] to facilitate the emptying of the bladder and to protect the kidneys. In addition, when the patient has a very limited bladder capacity, high intravesical pressure, and/or poor compliance, the patient may need a bladder augmentation.

Perhaps the most serious problem is the fact that many of these functional disorders, such as neurogenic bladder, may turn out to be progressive. This may result in a very unpleasant surprise later in life. Patients that behaved normally from the urinary function point of view may develop reflux and kidney damage. That is why all patients must be followed meticulously with emphasis in the preservation of the anatomy of the kidney, as well as its function.

We routinely recommend to do a kidney ultrasound 1 month postoperatively and subsequently 3, 6, and 12 months later and every year later, provided the patient has no urinary tract infections. When they have urinary tract infections, the patients deserve a full urologic evaluation.

23.14 Postoperative Problems

Many babies suffer from urologic problems consecutive to a damage provoked by a technically deficient operation performed to repair an anorectal malformation [79–81]. The most conspicuous problems that we have seen occurred in patients that were operated without a high-pressure distal colostogram (see Chap. 6, Sect. 6.6). We insist that the high-pressure distal colostogram is the most important diagnostic preoperative study in anorectal malformations, which allows us to determine the precise location of the recto-urinary fistula as well as the length of the bowel available from the colostomy to the fistula site. This information is vital for the surgeon to make a precise surgical plan and to avoid damage to important urologic structures. Approaching the patient posterior sagittally, looking for a rectum that is not there, frequently ends up in severe damage to other important structures. We have seen patients that suffer from urethral strictures (18 cases) and resections of the vas deferens, seminal vesicles, and prostate as demonstrated by the fact that the patients do not have ejaculation later in life. Also, the search for the rectum through a posterior

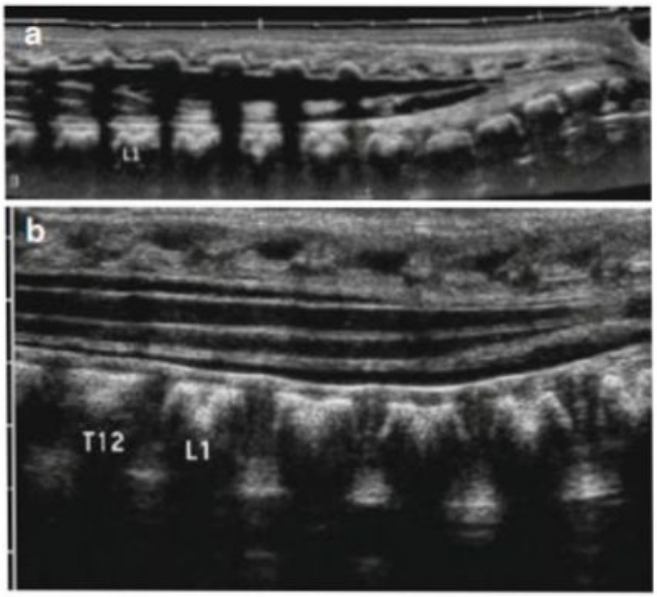

Fig. 23.17 Spinal Ultrasound. (a) Normal location of the conus. (b) Tethered cord

evidence of tethered cord, the patient has a normal sacrum, and in anorectal malformation considered "benign" in terms functional prognosis, we do not think that it is indicated to do an MRI of the spine, provided the patient has bowel and urinary control, as well as no lower extremity motion problems. On the other hand, if the ultrasound showed evidence of tethered cord, or is considered borderline or doubtful, we performed an MRI to confirm or to rule out the diagnosis. If the patient is asymptomatic and has a "good" malformation, we keep waiting until the age when the patient is willing to cooperate and remain quiet inside the rather scary and noisy MRI machine. We try to avoid the administration of heavy sedation or anesthesia as much as possible.

A good, scientifically oriented study is required to determine the real significance of the presence of tethered cord, as well as the indications for operation, benefits, and potential negative implications.

Tethered cord is frequently associated with malformations with very bad prognosis. Therefore, many of those patients have no bowel control and no urinary control, but we do not know how much is because of the tethered cord and how much is because of the poor sacrum and other negative anatomic features.

23.18 The Ultimate Concern, Kidney Function

Some patients with anorectal malformations are born with severe kidney damage, including end-stage renal disease [96]. Those patients represent one of the most serious therapeutic challenges. They must be treated by a multidisciplinary team in a sophisticated tertiary center, with experienced pediatric nephrologist and pediatric transplant surgeons [97]. Fortunately, the number of this type of cases is limited.

There is however a much larger and important group of patients who were born with relatively "good kidneys," but unfortunately they were not followed and monitored closely. The urologic problem (reflux, obstruction, infection, poor bladder function) was not treated adequately which resulted in severe kidney damage. This is preventable and must not happen.

We cannot over-empathize the importance of the frequent monitoring and long-term follow-up of patients with anorectal malformations [98]. This is particularly true in patients considered at risk of suffering kidney damage. These include cloacas [99, 100], bladder neck fistula, deficient sacrum, tethered cord, patients with hydronephrosis at birth, and patients with a single kidney.

In summary, urologic malformations are extremely important in patients with anorectal malformations. Pediatric surgeons should become aware of the fact that most of these patients have urinary problems and must become familiar with the way to detect and manage these problems, as well as to work together with a dedicated pediatric urologist in studying these kinds of problems. Also, we want to emphasize the importance of follow-up of these patients for life, to avoid damage and deterioration of the renal function.

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