Associated Genitourinary Tract Anomalies in Anorectal Malformations : A Thirteen Year Review

SURASAK SANGKHATHAT, M.D.*,
SAKDA PATRAPINYOKUL, M.D.*,
KAMOLNATE TADTAYATHIKOM, M.D.*

Abstract

Congenital anomalies in the genitourinary tract are the leading associated anomalies in infants with anorectal malformations (ARM). Certain anomalies such as vesicoureteric reflux (VUR) may cause permanent renal damage.

Objective : To review associated genitourinary tract anomalies in cases of anorectal malformations and evaluate the efficacy of ultrasonography in detecting VUR.

Material and Method : Retrospective review of 183 patients with ARM undergoing anoplasty between 1988 - 2001.

Results : Genital anomalies were found in 14 per cent (26 cases). Urologic anomalies were detected in 25.6 per cent (47 cases), with a higher incidence in supralevator anomalies. The most common upper tract anomaly was hydronephrosis, which resolved spontaneously in most of them. VUR was found in 16 cases (21 refluxing units) or 20 per cent of patients to whom voiding cystourethrogram (VCUG) was done. Sonography detected hydroureter and/or hydronephrosis in 3 of 21 refluxing units, despite 17 of them being grade three or more. Half of the cases with reflux had urinary tract infection at least once in the follow-up period despite normal initial urinalysis. Parenchymal scar was positive in four cases with VUR.

Conclusion : Thorough evaluation of the urinary tract is necessary in infants with anorectal malformations. Ultrasound is an accurate tool in the examination of the upper tract, but not sensitive enough to detect lower tract anomalies, especially VUR.

Key word : Anorectal Malformations, Genitourinary Tract Anomaly, Vesicoureteric Reflux

**SANGKHATHAT S,
PATRAPINYOKUL S, TADTAYATHIKOM K
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* Pediatric Surgery Unit, Department of Surgery, Faculty of Medicine, Prince of Songkla University, Songkhla 90110, Thailand.

Congenital defects in the genitourinary system are the leading associated anomalies in infants with anorectal malformations (ARM)[1-3]. The defects comprise both anatomical and functional derangement along the entire course of the urinary tract as well as the external genitalia. Functional obstructive anomalies such as the vesicoureteric reflux lead to recurrent urinary tract infection, which is the potential cause of permanent renal damage[4,5].

Screening for associated urologic anomalies needs radiologic investigation which varies among institutes[6]. Most case series do ultrasonography or intravenous pyelogram (IVP) to evaluate the anatomy of the upper urinary tract. Voiding cystourethrogram (VCUG) was advocated by Smith ED as a routine evaluation of lower tract in infants with anorectal malformations[4]. However, some protocols reserve VCUG for cases with urinary tract infection who have abnormal ultrasonographic study or parenchymal nuclear scan[7,8]. Moreover, thorough investigations are often neglected in patients with low type anomalies because it is believed that the incidence of associated anomalies is not significant[9].

This study aimed to review the associated genitourinary tract anomalies in infants with anorectal malformations and evaluate the efficacy of urinalysis and ultrasonography in detecting vesicoureteric reflux. The data are valuable in the establishment of practice guidelines for this group of patients.

MATERIAL AND METHOD

Medical records of 183 patients with anorectal malformations undergoing anoplasty in Songklanagarind Hospital between April 1988 and April 2001 were retrieved and reviewed regarding the type of malformation, evidence of associated genitourinary anomalies, their management and follow-up. The anorectal malformations were classified according to the Wingspread scheme (Table 1). Five cases of cloaca, one rectal duplication, two anovestibular fistula without imperforate anus and seven re-operative cases in whom the primary type could not be specified were also included.

For analysis, high and intermediate anomalies, cloacal malformations and rectal duplication were grouped together as the 'supralevator' group, whereas, low type anomalies and cases of anovestibular fistula without imperforate anus were grouped as the 'infralevator' group. The majority of the

Table 1. Type of anorectal malformations according to Wingspread classification. (N = 183)

Male anomalies	Cases	%	Female anomalies	Cases	%
High					
Anorectal agenesis with recto-vesical fistula	5	2.7			
with rectoprosthetic urethral fistula	8	4.4			
without fistula	5	2.7			
Intermediate			Intermediate		
Rectobalbar urethral fistula	37	20.2	Rectovestibular fistula	6	3.3
Anal agenesis without fistula	28	15.3	Rectovaginal fistula	12	6.6
			Anal agenesis without fistula	12	6.6
Low			Low		
Anocutaneous fistula	17	9.3	Anovestibular fistula	26	14.2
Anal stenosis	1	0.5	Anocutaneous fistula	6	3.3
			Anal stenosis (all with anterior ectopic anus)	5	2.7
Unspecified (redo-operation)	3		Cloacal malformations	5	2.7
			Isolated anovestibular fistula without imperforate anus	2	
			Rectal duplication	1	
			Unspecified (redo-operation)	4	
	104	56.8		79	43.2

Table 2. Associated inguino-scrotal and genital organs anomalies.

Genital anomalies	Supralevator group	Infralevator group	Unspecified type
Posterior hypospadias	7		
Bifid scortum	5		
Anterior hypospadias	2	1	
Micropenis	3		
Inguinal hernia	2 (1 bilateral hernia)		
Undescended testis	2 (2 bilateral UD)		1
Double vagina	1		1
Double cervix	1		
Labial hypertrophy	1		
Uterine agenesis	1		
Perineal hamartoma		1	

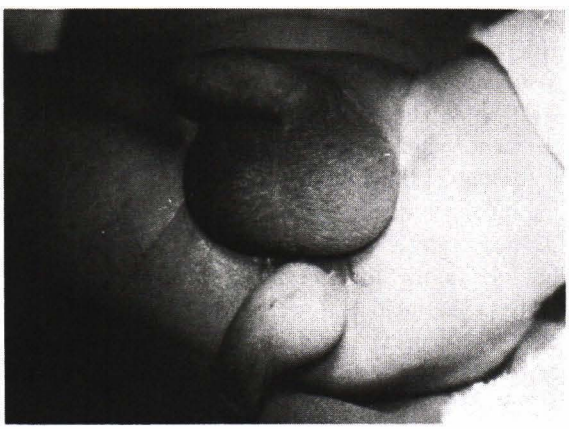

Fig. 1. Perineal hamartoma.

operations in the supralevator group were posterior sagittal anorectoplasty. Posterior myectomy and Y-V plasty plus anterior Z-plasty as described by Patrapinyokul[10] were performed in 40 of 57 cases of the infralevator group, with the remaining undergoing perineal anoplasty of other fashions.

Urinalysis was done in all patients. Ultrasonography of the urinary system was done in 149 patients (81.4%), which included all cases of cloaca and rectal duplication, 103 of 113 cases with high or intermediate types (91.2% of supralevator group), 36 of 57 cases with infralevator type malformations (63.2%) and 5 of 7 cases of unspecified type. VCUG was investigated in 79 patients (43.2%), comprising 64 cases (54.6%) and 12 cases (21.0%) in the supralevator and infralevator group, respectively. The severity of vesicoureteric reflux (VUR) was graded according to the International Reflux Study Committee. Vaginocystoscopy was performed in all patients with cloacal malformation. Mean follow-up period was 16.5 months (0 - 103 months). One case in the infralevator group died of cyanotic cardiac anomalies.

RESULTS

Demographic data

There were 183 patients, 105 males and 78 females, who underwent anoplasty during the thirteen-year period, ranging in age from one day to twenty years, with a median age of 7.0 months. Average weight was 7.8 kilograms (0.9-41 kg) One hundred and nineteen cases were classified as supralevator anomalies and 57 cases as infralevator. The overall incidence of associated genitourinary tract anomalies was 59 cases (32.2%). Associated anomalies other than the genitourinary systems were detected in 58 cases in the supralevator group (48.7%), 12 cases in the infralevator group (21.0%) and 2 of the 7 unspecified. The majority belonged to cardiopulmonary (31 cases), musculoskeletal (16 cases), vertebral system (14 cases) and esophageal atresia (7 cases). Thirteen patients (7.1%) had trisomy21 and 29 patients (15.8%) were in VACTERL association (three or more features).

Genital anomalies

Malformations of the genital tract and inguino-scrotal region occurred in 26 cases, the most common being posterior hypospadias (Table 2). Twenty-two cases in the supralevator group had associated genital anomalies, compared to only two in the infralevator group. Perineal hamartoma in a case of infralevator type is shown in Fig. 1.

Urologic anomalies

Urinalyses were abnormal in 28 cases of the supralevator type (23.5%) and 8 cases of the infralevator type (14.0%). Abnormal urologic screening by ultrasonography and/or voiding cystourethrogram was found in 47 cases (25.6%). The most common abnormal sonographic findings were hydronephrosis (Table 3). Among 11 cases with absent renal shadow, a single kidney was confirmed by intravenous pyelogram in all cases, including one patient with pelvic-single right kidney in whom ultrasonography failed to detect any renal echo. IVP in cases of single kidney also demonstrated four cases of duplex kidneys and three crossed-ectopia, while a sonogram detected only three duplexes and no ectopia. Voiding cystourethrogram revealed vesicoureteric reflux in 16 patients. Marked trabeculated bladder mucosa with or without diverticulum, suggestive of spastic neurogenic bladder, were noted in 9 cases.

The majority of patients with hydronephrosis found by ultrasound were asymptomatic and had spontaneous resolution after anoplasty procedures. Among 26 renal units with hydronephrosis, only three had vesicoureteric reflux. Three of six patients with bilateral hydronephrosis had mucosal trabeculation and diverticular bladder.

Vesicoureteric refluxes detected in 16 patients included 9 left sided, 2 right sided and 5 bilateral refluxes or a total of 21 refluxing units. Incidence of reflux in the supralevator type was 12 of 64 cases in whom VCUG had been performed (18.7%) and 3 of 11 cases in the infralevator group (27%). Surprisingly, ultrasonography detected hydronephrosis and/or hydroureter in only three refluxing units (14.3%), despite 17 of 21 being grade 3 or more. Eight of 16 patients with VUR experienced urinary tract infection at least once during their follow-up period, although initial urinalyses were unremarkable in 10 of 16 cases. Renal parenchymal scan (DMSA) was done in 5 patients and the studies revealed perenchymal scar in four cases. Clinical symptoms and/or radiologic signs of spastic bladder were associated in six patients, all of them had dilated ureter (grade 3 or more) with bilaterality in three patients. Two of sixteen cases with VUR had caudal regression syndrome. (Fig. 2)

Fourteen cases of VUR were managed conservatively with oral prophylaxis antibiotics and serial evaluation by VCUG or renal scan. All VUR less than grade 2 resolved spontaneously. One case with bilateral reflux who also had neurogenic bladder developed hypertension. No patients had azotemia. A case of left single kidney and severe reflux underwent ureteroneocystostomy.

DISCUSSION

Associated genitourinary tract anomalies have been reported in 26 to 59 per cent of pediatric patients with anorectal malformations[1-6]. The incidence varied with the level of the rectal pouch, the aggressiveness of the screening protocol, and the definition of "anomalies". High type anorectal malformations usually had a higher incidence. Pena A. found 78 per cent of urological anomalies in ARM with recto-bladder neck fistula compared to 28 per cent in the cases with perineal fistula[11]. Hoekstra WJ reported 47 per cent and 35 per cent association in supralevator and infralevator, respectively[12]. The authors' overall incidence of 32.2 per cent, which were 38.6 per cent in the supralevator and 21.0 per cent in the infralevator, supports the influence of rectal pouch level on the incidence. A lower rate of thorough evaluation may be attributable to the low incidence of associated anomalies. The incidence in the infralevator group was also less than 31 per cent in a series reported by Mistra D et al[9]. The corrected incidence of VUR at 20 per cent is comparable with that reported by Parrott TS (19%)[13] and Boemers TML (27%)[14], however, it is approximately half of the reports of Narasimharao KL (47%)[3], and Rickwood AMK (45%)[15]. The incidence of hypospadias, bifid scortum and undescended testis did not differ from other series[12,13].

The ultrasonogram is an appropriate diagnostic tool in detecting structural anomalies of the upper urinary tract. The study was recommended to replace IVP which was commonly performed in the past[16]. According to the authors' experience, ultrasound has some limitations in detecting misplaced or duplex kidneys. Moreover, in the majority cases with vesicoureteric reflux, ultrasound results were negative, while in contrast, reflux was demonstrated in only a few cases with hydronephrosis detected by ultrasonogram. A large series of pediatric patients with urinary tract infection, stated that 23 per cent of patients who had negative ultrasonogram and DMSA scans showed VUR on VCUG. Even severe reflux may have no dilatation noted on the ultrasound study[18]. The presented data and other literature suggests that an ultrasonogram alone

Table 3. Associated urologic anomalies found by ultrasonography and voiding cystourethrogram.

Group of anorectal malformations	Abnormal urinalysis (cases)	%	Ultrasound findings		Voiding cystourethrogram	
Supralevator	28	23.5	Unilateral absence of kidney	9**	Vesicoureteric refluxes	
			Hydronephrosis		Unilateral	8
			Unilateral	9	Bilateral	4
			Bilateral	5	Mucosal trabeculation*	7
			Malrotation of kidney	2	Double collecting system (duplex kidney)	1
			Multicystic kidney	1	Bicornuate bladder	1
			Duplex kidney	3		
Infralevator	8	14.0	Unilateral absent of kidney	1**	Vesicoureteric refluxes	
			Hydronephrosis, unilateral	3	Unilateral	3
			Non-visualization of both kidneys	1***	Bilateral	1
			Cystic kidney	1	Mucosal trabeculation*	1
			Malrotation of kidney	1		
Unspecified type	0		Hydronephrosis		Mucosal trabeculation*	1
			Unilateral	2		
			Bilateral	1		

* without bladder outlet obstruction

** all renal agenesis were comfirmed by intravenous pyelogram (IVP), 3 of the 8 cases also had crossed ectopia

*** IVP showed duplex-pelvic right kidney

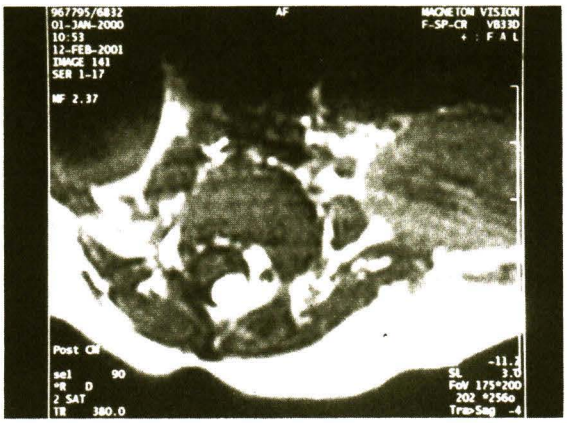

Fig. 2. Magnetic resonance imaging of spine, showing dural lipoma in an infant with high type anorectal malformations, sacral agenesis and bilateral severe vesicoureteric reflux.

is not an adequate screening tool in evaluation of the urinary system in patients with ARM, whose major problems are functional and situated in the lower urinary tract.

Voiding cystourethrogram is a standard radiologic study in detecting VUR. The contour and alignment of mucosa are also deliniated[19]. Although the incidence of neurogenic bladder cannot be concluded from the present data, six of sixteen cases with VUR manifested symptoms of neurogenic bladder (incontinence) and two cases also had caudal regression syndrome. Urodynamics studied revealed correlation between spine anomalies, neurovesical dysfunction and the development of VUR in ARM patients[20]. Recent urodynamic study in 90 children with ARM found that 21 of 38 patients with sacral anomalies had neurogenic bladder, 60 per cent of them had VUR and half suffered from reflux nephropathy[14,21]. However, the benefit of neurosurgical treatment for a child with neurogenic bladder remains a debatable issue.

Death from renal failure was reported to be as high as 6 per cent of ARM patients[2]. Four of 29 patients with low type ARM together with urologic anomalies, in a review by Mistra D, had evidence of chronic renal insufficiency[9]. Although in the present series there was no mortality from renal failure, four cases had parenchymal scar. Two of them were in the infralevator group and one developed secondary hypertension. The data suggest that certain structural and genitourinary tract anomalies associated with anorectal malformations, especially VUR, predisposes this group of patients to significant morbidity. Urinalysis and ultrasonography are not sensitive enough for early detection of VUR. Complete urinary tract assessment should be composed of upper tract imaging by ultrasonogram, together with lower tract study by VCUG, regardless of the types of anorectal malformations. Early detection may reduce the occurrence of urinary tract infection as well as the risk of permanent renal damage.

SUMMARY

The authors retrospectively reviewed associated urogenital anomalies in pediatric patients with anorectal malformations and emphasized the necessity of thorough evaluation in all types.

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ความพิการร่วมของระบบทางเดินปัสสาวะและอวัยวะสืบพันธุ์ในผู้ป่วยที่มีความผิดรูปโดยกำเนิดของทวารหนักและไส้ตรง

สุรศักดิ์ สังขทัต ณ อยุธยา, พ.บ.,*
ศักดา ภัทรภิญโญกุล, พ.บ., กมลเนตร ตัถยาธิคม, พ.บ.**

ได้ทบทวนอุบัติการณ์ของความพิการร่วมของระบบทางเดินปัสสาวะและอวัยวะสืบพันธุ์ในผู้ป่วยเด็กที่มีความผิดรูปโดยกำเนิดของทวารหนักและไส้ตรง 183 รายซึ่งเข้ารับการผ่าตัดระหว่างเดือนเมษายน พ.ศ. 2531 ถึงเดือนเมษายน พ.ศ. 2544 และประเมินประสิทธิภาพของเครื่องมือวินิจฉัยต่าง ๆ ได้แก่การตรวจปัสสาวะ การตรวจอัลตร้าซาวด์ ในการตรวจกรองความพิการร่วม พบผู้ป่วยมีความพิการร่วมของอวัยวะสืบพันธุ์ร้อยละ 14 (26 ราย) ความพิการร่วมของระบบทางเดินปัสสาวะร้อยละ 25.6 (47 ราย) ความพิการร่วมที่พบมากที่สุดของทางเดินปัสสาวะส่วนบนคือไตโป่ง ซึ่งส่วนใหญ่หายได้เองหลังจากที่ผ่าตัดซ่อมสร้างทวารหนักแล้ว พบการไหลย้อนของปัสสาวะจากกระเพาะปัสสาวะขึ้นสู่หลอดไต 21 ข้างในผู้ป่วย 16 ราย ในจำนวนนี้เพียง 3 ข้างสามารถตรวจพบไตหรือหลอดไตโตโดยการทำอัลตร้าซาวด์ ผู้ป่วยที่มีการไหลย้อนครึ่งหนึ่งมีการติดเชื้อของทางเดินปัสสาวะอย่างน้อยหนึ่งครั้งในระหว่างที่ติดตามการรักษา พบมีความผิดปกติของเนื้อไตในผู้ป่วยที่มีการไหลย้อน 4 ใน 5 รายซึ่งได้รับการตรวจสแกนเนื้อไต กล่าวโดยสรุป ความพิการร่วมของระบบทางเดินปัสสาวะและอวัยวะสืบพันธุ์พบได้บ่อยในทารกที่มีความพิการโดยกำเนิดของทวารหนักและไส้ตรง ความพิการร่วมดังกล่าวควรได้รับการตรวจหาโดยการทำอัลตร้าซาวด์เพื่อประเมินทางเดินปัสสาวะส่วนบนและตรวจ voiding cystourethrogram เพื่อประเมินทางเดินปัสสาวะส่วนล่าง

คำสำคัญ : ความผิดรูปโดยกำเนิดของทวารหนักและไส้ตรง, ความพิการร่วม, การไหลย้อนของปัสสาวะ

สุรศักดิ์ สังขทัต ณ อยุธยา,
ศักดา ภัทรภิญโญกุล, กมลเนตร ตัถยาธิคม
จดหมายเหตุทางแพทย์ ฯ 2545; 85: 289-296

* หน่วยกุมารศัลยศาสตร์, ภาควิชาศัลยศาสตร์, คณะแพทยศาสตร์ มหาวิทยาลัยสงขลานครินทร์, สงขลา 90110