Associated Genitourinary Abnormalities in Low-type Anorectal Malformation and Urological Investigations

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Objective: *Urogenital anomaly has been considered as the most common associated condition with anorectal malformation (ARM). There were recommendations for urogenital anomaly surveillance. The goal of this study was to evaluate the benefit of screening tools for urogenital anomalies in ARM patients, especially low type malformations, in our institution.*
Material and Method: *A retrospective review of 183 ARM patients in Siriraj Hospital between January 2004 and December 2014 was performed. Demographic data included age, sex, type of anorectal malformation, type of surgery, and other associated anomalies. Basic screening methods are ultrasonography and voiding cystourethrography. Further investigations included diuretic renogram, intravenous pyelography, radionuclide scan, magnetic resonance urography, cystoscopy, and retrograde pyelography. Symptoms, types of anomaly, and treatments of urinary tract anomaly were recorded.*
Results: *All 183 patients were reviewed. Low type malformation accounted for 51 patients. One hundred and fifty three were scheduled for renal ultrasonography as a screening protocol, in which 42 had abnormal results. The most common anomaly was vesicoureteric reflux followed by renal agenesis. Thirteen low-type ARM patients had abnormal screening results. After follow-up, 4 out of 11 hydronephrotic and pelviectatic patients appeared normal in later years. Hypospadias was the predominated genital abnormality among low type ARM with urinary tract anomaly patients. This correlation was not found in non-low type patients.*
Conclusion: *Hydronephrosis was the most common genitourinary abnormality in low type ARM, which some of them spontaneously resolved. Expectant management was preserved in selected cases with normal kidney function. Noninvasive screening tests should be encouraged and performed in all ARM patients regardless of the types.*

Keywords: *Anorectal malformation, Genitourinary abnormality*

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Anorectal malformation (ARM) has high incidence of associated anomalies, varying from 50% to 75%. Urogenital anomaly has been concerned as the most common associated condition to anorectal malformation[1], with varying incidences ranging from one third to a half in some series[2,3]. The more complex malformation of the anus, the more urogenital anomaly was found[4]. Recommendations for associated urogenital anomaly screening in anorectal malformation patients were initiated and universally conducted. The screening programs were helpful in terms of early detection and long term follow-up in urogenital anomaly patients[5]. However, in developing countries, insufficient resource is still the main problem in improving patient's care. To detect urogenital anomalies, various tools are involved. In Siriraj Hospital, a screening investigation for urogenital anomalies inARM patients included spinal and kidney ultrasonography. Some low type ARM patients were missed from the protocol, although they had no urogenital problem later in life. Therefore, the benefit of routine investigations for detecting associated urogenital anomalies in low type anorectal malformation is questionable. The goal of this study was to evaluate the benefit of screening tools for urogenital anomalies in ARM patients, especially low type malformations.

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Material and Method

A retrospective review of patients presented with anorectal malformation and underwent definitive surgery in Siriraj Hospital between January 2004 and December 2014 was conducted. One hundred and eighty three patients were included into the study.

Demographic data obtained included age, sex, type of anorectal malformation, type of surgery, and other associated anomalies of the patients.

The types of Anorectal malformation were classified by Krickenbeck Classification[6]. Low type ARM was described as either with cutaneous (perineal) fistula or without fistula when the rectal pouch reaching less than 1 cm from skin. For low type anorectal malformation, anoplasty could be performed immediately at birth. Non-low type anorectal malformation referred to other ARM including complex anomalies with the rectal pouch locating more than 1 cm from skin.

Basic screening methods for urinary tract anomaly were ultrasonography and voiding cystourethrography. Further investigations included diuretic renography, intravenous pyelography, DMSA (Dimercaptosuccinic acid) radionuclide scan, magnetic resonance urography, cystoscopy, and retrograde pyelography. Symptoms and treatments of urinary tract anomaly were recorded.

Concerning urinary tract anomalies included renal agenesis, multicystic dysplastic kidney disease, hydronephrosis, hydroureter, ureteric duplication, ectopic ureterocele, ureteropelvic junction obstruction, ureterovesical junction obstruction, horse shoe kidney, neurogenic bladder, vesicoureteric reflux and posterior urethral valve. All data were obtained from Siriraj Hospital patient databases.

The correlation between genital abnormality such as hypospadias, micropenis, undescended testis, indirect inguinal hernia and types of anorectal malformation were also studied.

Results

There were183 patients included into the study. One hundred and seven patients were male and 76 were female. The types of ARM were categorized in Table 1. Low type malformation accounted for 51 patients (37 male and 14 female) (Table 1).

Of the 183 patients included, 153 were scheduled for KUB (kidneys, ureters and bladder) ultrasonography as a screening protocol. Among the 30 patients who did not have ultrasonography, only 1 developed clinical urinary tract infection which was caused by urinary reflux from a long common channel in persistent cloaca. Others had no urinary tract symptom.

Of the 153 screened patients, 101 patients had normal results from KUB ultrasonography. There were 27 low type anorectal malformation patients and 74 non-low type patients. Comparison of the results of the urinary tract anomaly screening between the types of anorectal malformation, was demonstrated in Table 2.

One patient with low type anorectal malformation, who had normal KUB ultrasonography from initial screening test, later developed urinary tract infection. Further investigation was suggestive of neurogenic bladder, which suprapubic cystostomy was conducted.

Patients who had abnormal results from screening KUB ultrasonography obtained various further investigations, depending on findings detected from KUB ultrasonography. Investigation tests included voiding cystourethrography, intravenous pyelogram, cystoscopy, DMSA scan, diuretic renogram, and magnetic resonance urography. The most common urinary tract anomaly found in this review was vesicoureteric reflux (13 patients) followed by renal

Table 1. Associated urinary tract anomaly patients classified by types of anorectal malformation

	No. of patients (n = 107)	Urinary tract anomaly patients (%)		No. of patients (n = 76)	Urinary tract anomaly patients (%)
Male	**107**	**33 (30.8)**	**Female**	**76**	**15 (19.7)**
Cutaneous (perineal) fistula	28	2 (7)	Cutaneous (perineal) fistula	12	0
Rectobulbar fistula	25	5 (20)	Vestibular fistula	46	9 (19.5)
Rectoprostatic fistula	14	7 (50)	Imperforate anus without fistula (low type)	2	2 (100)
Recto-Bladder neck fistula	13	7 (53.8)	Imperforate anus without fistula (non-low type)	9	0
Imperforate anus without fistula (low type)	9	5 (55)	Rectal atresia	0	0
Imperforate anus without fistula (non-low type)	18	5 (27.7)	Cloaca	7	4 (57)
Rectal atresia	0	0			

agenesis (11 patients). The remaining were hydronephrosis, horseshoe kidney, multicystic dysplastic kidney disease, neurogenic bladder, double collecting system and renal hypoplasia.

Thirteen low type anorectal malformation patients had abnormal hydronephrosis demonstrated from screening test, in which follow-up ultrasongraphy and further investigations showed resolution and normal results in 4 patients. Seven patients with history of hydronephrosis showed no symptoms of urinary tract obstruction or infection. One patient with renal tubular acidosis was found having bilateral nephrocalcitosis as demonstrated in Table 3.

More complex anomalies of the urinary tract were identifiedin non-low type malformation. Vesicoureteric reflux and renal agenesis were predominated in this group. Urinary tract anomalies in non-low type anorectal malformation patients were demonstrated in Table 3.

In the non-low typeanorectal malformation group, there were 5 patients who had combined anomalies. A multicystic dysplastic kidney disease patient had hydro nephrosis correlated with posterior urethral valve. Two neurogenic bladder patients had horseshoe kidney in association with vesicoureteric reflux, whereas 2 renal agenesis patients had vesicoureteric reflux in the contralateral functioning kidney.

The incidences of associated urinary tract anomaly in the study group, classified by the types of ARM, were demonstrated in Table 1. Urinary tract anomalies were noticeably more observed in more complex type of ARM patients. The results are consistent with findings from other series[3,7].

The associated genital anomalies including hypospadias, micropenis, undescended testicle and indirect inguinal hernia were studied in correlation with the types of anorectal malformation. The results were demonstrated in Table 4. Interestingly, all low type ARM patients who had associated genital anomalies also demonstrated abnormal upper urinary tract structure. None of the patients in low type ARM group with abnormal genitalia had normal upper urinary tract.It could be hypothesized from these results that urinary tract abnormality should be suspected in any low type ARM patient who demonstrated genital anomaly.

Table 2. Comparison of the urinary tract anomaly screening between types of anorectal malformation

	Low type n = 51 (%)	Non-low type n = 132 (%)
Screening for urinary tract anomaly	40 (79.5)	113 (85.6)
Not screened	11 (21.5)	19 (14.4)
Normal results	27 (67.5)	74 (65.5)
Abnormal results from screening	13 (32.5)	39 (34.5)

Table 3. Urinary tract anomalies in Low and Non-low type anorectal malformation patients

Urinary tract anomaly	Male		Female	
	Low-type ARM	Non-low type ARM	Low-type ARM	Non-low type ARM
Renal agenesis	0	6	0	5
Multicystic dysplastic kidney disease	0	3	0	1
Hydronephrosis	7	4	0	0
Ureteropelvic junction obstruction	0	1	0	0
Horseshoe kidney	1	4	0	0
Neurogenic bladder	0	2	1	0
Vesicoureteric reflux	0	8	0	5
Posterior urethral valve	0	2	0	0
Renal hypoplasia	0	0	0	1
Double collecting system	0	1	0	1
Bilateral medullary nephrocalcitosis	0	0	1	0

Table 4. Genital anomalies in anorectal malformation patients

Genital abnormality	Low type anorectal malformation		Non-low type anorectal malformation	
	Normal upper urinary tract	Abnormal upper urinary tract	Normal upper urinary tract	Abnormal upper urinary tract
Hypospadias	-	3	2	1
Undescended testis	-	1	2	2
Micropenis	-	1	-	-
Indirect inguinal hernia and hydrocele	-	-	2	-

Discussion

In the present study, data of patients with ARM in Siriraj Hospital over 10 consecutive years from January 2004 to December 2014 were collected and analyzed. Of the 183 patients included, 107 were male predominating (58.5%). The incidence of low-type ARM was 37 out of 107 male patients and 14 out of 76 female, accounting for 34.5% and 18.4% respectively. Vestibular fistula was the most common type of ARM in female at 60.5% (n = 46), all of which could be corrected at birth by anal transposition procedure.

Ultrasonography of the KUB system was indicated in all patients with ARM treated in Siriraj Hospital however there were 30 patients (16.3%) missed from the protocol. Among these, cutaneous fistula and vestibular fistula were the most frequently missed groups. Considering these 2 groups mostly had single hospitalization and ultrasonography was scheduled later, therefore some patients were missed from the screening test.

According to Table 1, the incidence of urinary tract anomaly classified by types of ARM, urinary tract anomalies were noticeably more observed in more complex type of ARM patients. The resultis consistent with findings from other series[3,7]. However, in low-type ARM patients, there were at least 18.9% male and 14.3% female patients suffering from associated urinary tract anomaly. Those who were missed from the screening protocol and remained asymptomatic were not included. Considering that at least 1 out of 5 low-type ARM patients have associated urinary anomaly, screening tests for urinary anomaly remain important for this group.

Hydronephrosis was the most common KUB anomaly associated with low-type ARM in the screening test. There were 4 out of 13 patients who had either hydronephrosis or pelviectasia, later found to have normal results after observation and further investigations. Interestingly, these findings were also observed in other series[5] which expectant management was used in those who had no symptoms and good renal function. Accordingly, asymptomatic and good kidney's function detected by ultrasonographic screening could be managed by close observation and regular following-up.

More complex urinary tract anomalies were observed in higher and more complicated ARM patients. These urinary tract anomalies mostly required subsequent medical or surgical treatment. In cases of renal agenesis, patients and families should be informed for awareness of single kidney. Furthermore, other associated anomalies such as vesicourinary reflux could worsen long term prognosis in these patients. Similar findings were mentioned in other studies which also emphasize the importance of screening test in non-low type anorectal malformation[8].

Regarding associated genital abnormality, hypospadias was predominated in low type ARM with urinary tract anomaly. Interestingly all low type ARM patients who had genital anomaly demonstrated associated upper urinary tract abnormality as well. Although this correlation was not consistent with non-low type ARM, we believe that urinary tract abnormality should be suspected in low type ARM associated with genital abnormality.

Conclusion

More complex ARM was associated with a higher risk of having urogenital abnormalities. Hydronephrosis was the most common genitourinary abnormality in low type ARM patients. Screening tests for urinary anomaly is still important for this group. Expectant management was preserved in selected cases with normal kidney function. Noninvasive screening tests should be performed in all ARM patients. Further investigation should be conducted for patients who had abnormal screening test results. Tools for further investigation should be selected accordingly.

What is already known on this topic?

Urological anomalies are the most common associated anomaly of ARM and this has influenced establishment of screening protocol. At present, the ultrasonography of kidney is indicated in all ARM patients.

What this study adds?

This study showed data of urologic abnormality associated with ARM patients which were divided into low and non-low type group and found that the incidence was less in low-type patient. Furthermore, in some patient, the abnormality was spontaneously resolved. This encourage us on expectant management in selected patient.

This study focused on urological abnormalities in low type ARM patients, benefit of the screening tool, and management of each group.

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Potential conflicts of interest

None.

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ความผิดปรกติของระบบทางเดินปัสสาวะและอวัยวะเพศในทารกที่มีความผิดรูปของทวารหนักชนิดต่ำและการตรวจพิเศษเพิ่มเติมของระบบทางเดินปัสสาวะ

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วัตถุประสงค์: *ความผิดปรกติของระบบทางเดินปัสสาวะเป็นภาวะที่พบได้มากในผู้ที่มีความผิดรูปของทวารหนักโดยปัจจุบันมีการตรวจพิเศษเพิ่มเติมในทารกกลุ่มนี้เพื่อการวินิจฉัยและรักษาที่รวดเร็ว ในการวิจัยนี้มุ่งเน้นการศึกษาถึงการตรวจคัดกรอง และตรวจพิเศษเพิ่มเติมเกี่ยวกับความผิดปรกติของระบบทางเดินปัสสาวะในกลุ่มผู้ป่วยที่มีความผิดรูปของทวารหนักชนิดต่ำในโรงพยาบาลศิริราช*

วัสดุและวิธีการ: *การศึกษานี้ทำในผู้ป่วยที่มีความผิดรูปของทวารหนัก 183 คนระหว่างปี พ.ศ. 2547 ถึง 2557 โดยเก็บข้อมูลได้แก่ อายุ เพศ ประเภทของความผิดรูปของทวารหนัก การรักษา และความผิดปรกติอื่นๆ ที่เกี่ยวข้อง เครื่องมือที่ใช้ในการตรวจคัดกรองผู้ที่มีความผิดปรกติของระบบทางเดินปัสสาวะคืออัลตราซาวด์และการตรวจโดยการฉีดสารทึบรังสี ผู้ที่มีความผิดปรกติจะได้รับการตรวจเพิ่มเติมอื่นๆ และเก็บข้อมูลแยกชนิดของความผิดปรกติ อาการแสดงและการรักษา*

ผลการศึกษา: *ผู้ป่วยได้รับการคัดกรองด้วยการตรวจอัลตราซาวด์ระบบทางเดินปัสสาวะ 153 คน และมี 51 คน ที่มีความผิดรูปของทวารหนักชนิดต่ำ พบว่ามีผู้ป่วย 13 จาก 51 คนที่มีความผิดปรกติของระบบทางเดินปัสสาวะหลังจากได้รับการตรวจเพิ่มเติมและการติดตาม ผู้ป่วย 4 คน จาก 11 คน สามารถหายจากภาวะกรวยไตมีปัสสาวะขัง (Hydronephrosis) ที่ตรวจพบจากการคัดกรองได้เอง นอกจากนี้ยังพบว่าในผู้ป่วยที่มีความผิดปรกติของระบบสืบพันธ์ มักพบร่วมกับผู้ที่มีความผิดปรกติของระบบทางเดินปัสสาวะในกลุ่มผู้ที่มีความผิดรูปของทวารหนักชนิดต่ำอีกด้วย*

สรุป: *จากการศึกษาพบว่าในกลุ่มผู้ที่มีความผิดรูปของทวารหนักชนิดต่ำพบว่ามีภาวะกรวยไตมีปัสสาวะขังมากที่สุดในผู้ที่มีความผิดปรกติของระบบทางเดินปัสสาวะ ในบางกรณีสามารถหายได้เองจากการติดตาม นอกจากนี้การตรวจคัดกรองเช่น อัลตราซาวด์ระบบทางเดินปัสสาวะมีความสำคัญและควรได้รับการส่งเสริมในผู้ป่วยกลุ่มนี้*