

## Urogenital Tract Abnormalities Associated with Congenital Anorectal Malformations

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### Abstract

**Objective:** Genitourinary anomalies in patients with imperforate anus are a frequent source of significant morbidity. Variability of reports on the incidence of associated anomalies with imperforate anus mandates investigation on this issue.

**Material & Methods:** The case records and imaging studies of 105 patients who underwent surgery for imperforate anus over a 10-year period are retrospectively reviewed. Voiding cystourethrogram, intra venous pyelography (IVP) and ultrasound were performed in patients with intermediate or high level anorectal lesions.

**Findings:** During 10 years, there were 48 boys (45.7%) and 57 girls (54.3%) with imperforate anus. Genitourinary anomalies were seen in 34 (48.6%) patients with intermediate or high level anorectal lesions. Eighteen of them (52.9%) were shown to have vesicoureteral reflux, only 6 of them required surgical correction. Vesicoureteral reflux was the most prominent urologic anomaly; other anomalies such as ureteropelvic junction (UPJ) stenosis, hydronephrosis, hypospadias, renal agenesis and undescended testis were seen in these series.

**Conclusion:** Patients with anorectal malformations should be evaluated for urinary tract and spinal anomalies.

**Key Words:** Imperforate anus; Anorectal malformation; Urinary tract anomaly

### Introduction

Anorectal malformations are a complex group of malformations diagnosed at birth because of absent or ectopic anus<sup>[1]</sup>. There is

variable data, however the incidence is approximately 1:5000 in live births. Genitourinary anomalies

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occur frequently in the patients with congenital anorectal malformation. Urinary tract problems are common in these patients with a reported incidence of 26% to 50% in several large series<sup>[1,2]</sup>. Most of the genital anomalies are visible on clinical examination, but urological anomalies need investigations for their detection<sup>[3]</sup>.

The purpose of this study was to evaluate the frequency of coexisting urinary malformations in a single population of patients with imperforate anus in our children's medical center.

## Material & Methods

During 10 years, from 1996 till 2005 in Mofid Children's Hospital totally 105 children with congenital anorectal malformations retrospectively were studied. All patients with apparent diagnosis of imperforate anus were included in this study.

The patient's sex, anorectal lesion level and the presence of urinary tract, genital or spinal anomalies were recorded. Level of anorectal lesion was determined by radiographic evaluation.

Renal ultrasound or intra venous pyelography (IVP) and Voiding cystourethrogram (VCUG) were performed for urinary tract malformations in all cases with intermediate or high level anorectal lesions but not in low level anal lesion. In patients with low level imperforate anus, genital malformations were defined as any anomaly of the penis, testis or scrotum in boys and vagina, cervix or uterus in girls.

## Findings

During 10 years, there were 48 boys (45.7%) and 57 girls (54.3%) with imperforate anus. Of the 105 patients, 35 patients (34.3%) had low level lesions. Totally, 70 (66.6%) of patients had intermediate or high level anorectal lesions, that's included 38 boys

(54.2%) and 32 girls (45.7%). Ten boys (28.6%) and 25 girls (71.4%) girls had low anomalies.

In patients with intermediate or high level anorectal lesions renal ultrasound or excretory urography and VCUG was performed for urinary tract malformations. The incidence of urinary tract anomalies was 48.6% in patients with intermediate or high level anorectal lesions. The most frequently encountered lesion was vesicoureteral reflux (VUR) which affected 18 (25.7%) of the patients, 8 of whom had bilateral type of the lesion (table 1). One of these patients had bilateral scar kidneys with chronic renal failure. Twelve patients with VUR were under medical supervision and treatment for a period of 2 to 4 years, and only 6 patients required surgical correction, and others are treated medically.

**Table 1-** Anomalies of urinary tract

Anomaly type	Frequency
Reflux	18 (52.9%)
Renal agenesis	4 (11.8%)
Hydronephrosis	4 (11.8%)
Renal hypoplasia	3 (8.8%)
UPJ* stenosis	3 (8.8%)
Duplication	2 (5.9%)
Total	34 (100%)

\*Ureteropelvic junction

Genitalia anomalies were found only in male patients. The incidence of genitalia anomalies was 13 males (27.1%) boys. The most frequently encountered anomaly was undescended testis (table 2).

Lumbosacral anomalies were found only in high type lesion. Ten patients (9.5% all patients or 14.2% high type) had lumbosacral anomalies including sacral agenesis in 8 (7.6%) and spina bifida in 2 (1.9%) cases. Two of them developed neurogenic bladder.

**Table 2-** Anomalies of the genitalia

Anomaly type	Frequency
Undescended testis	7 (54%)
Hypospadias	3 (23%)
Bifid scrotum	3 (23%)
Total	13 (100%)

## Discussion

The association of genitourinary anomalies with imperforate anus has been recently reviewed. In general, incidence of associated genitourinary anomalies ranging from 26% to 50% has been reported with an increased incidence of high (supralevator) as compared to low (infralevator) lesions<sup>[1,3,4]</sup>. In our study, the incidence of urinary tract anomalies was 48.6% in intermediate or high level anorectal lesions, and an overall rate was 32.4%. The findings of this study are similar to urinary tract anomalies reported in other studies<sup>[1,2,5]</sup>. The incidence of urinary tract anomalies increased with a higher level of anorectal malformation<sup>[6]</sup>.

VUR and renal agenesis are the most common associated urinary tract anomalies with imperforate anus. In our studies, VUR was the most common anomalies in 25.7% patients with high lesion and 52.9% patients with urinary tract anomalies. VUR was reported the most common associated urinary tract anomaly with imperforate anus<sup>[7]</sup>. The incidence of VUR in patients with anorectal malformations was different in various studies form 19% to 47.2%<sup>[2,4,8]</sup>. The current practice is to perform ultrasound which can be used to image the kidneys, bladder and to evaluate the spinal cord for tethering<sup>[9]</sup>.

Metts and Boemers found VUR in 32% of their cases<sup>[7,8]</sup>. Misra et al reported that 7.5% of patients with low deformity had VUR<sup>[10]</sup>,

but Tohda and Moore reported the incidence of VUR only in 0.7% and 5.4% of their patients<sup>[6,11]</sup>. This wide variation in incidence of VUR is related to the different methods of study. In some studies VCUG was performed only when some graphic findings were abnormal<sup>[12]</sup>.

In other study, hydronephrosis and renal agenesis were the most common abnormalities of the upper urinary tract, and neurovesical dysfunction is a frequent finding in children with anorectal malformations<sup>[13]</sup>. We found hydronephrosis and renal agenesis as two common of the upper urinary tract abnormalities. Neuro-vesical dysfunction commonly is associated with sacrospinal deformities. Some authors recommend evaluation of all patients with MRI, because spinal cord anomalies may occur without obvious sacrospinal anomalies<sup>[9]</sup>. Urodynamic studies (UDS) are reserved for those children with either a deformity of the spine or a spinal cord defect<sup>[13]</sup>.

Spine anomalies in other studies reported between 16-27%<sup>[2,6]</sup>, but in our series were 14.2%. In our patients, cryptorchidism and was the most common genital anomalies<sup>[14]</sup>.

## Conclusion

All patients with imperforate anus should be investigated for urogenital and spinal anomalies. Every effort should be unetaken to detect the associated urogenital anomalies, so that a better outcome can be expected in anorectal malformation. There is also an intense need to search for predisposing factors responsible for associated anomalies.

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