

## Genitourinary Anomalies in Patients with Anorectal Malformation

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### ABSTRACT

#### Objective

To determine the frequency of genitourinary abnormalities in patients having anorectal malformations.

#### Methodology

This descriptive cross sectional study was carried out in department of Pediatric Surgery, Khyber Teaching Hospital, Peshawar from August 2017 To February 2018. Any child (both genders) with clinically apparent diagnosis of anorectal malformation having age 1 month or less (Neonate) was included in the study. Any patient who has undergone any surgical intervention elsewhere for anorectal malformation and cases of intersex or genital ambiguity were excluded. Level of anorectal lesion was determined by radiographic evaluation. Renal ultrasound and intravenous-pyelography (IVP) and Voiding Cysto-urethrography (VCUG) were performed for urinary tract malformations in all cases with high level anorectal lesions. In low level anorectal lesion ultrasound was performed initially with IVP and VCUG performed later if needed to investigate any anomaly (dilated ureters, hydronephrosis) found on ultrasound.

#### Results

Total of 90 patients were included in this study. There were 68.89% males and females were 31.11%. The male to female ratio was 2.2:1. Average age of the patients was 5.03±6.11 days. The frequency of genitourinary abnormalities among children with anorectal malformations was found in 56(62.82%) patients with vesicoureteric reflux (VUR) being the most common anomaly (23.3%).

#### Conclusion

Anorectal Malformation patients should be screened for urogenital anomalies. Early detection of the associated anomalies will result in proper management and better outcome.

#### Key words

Anorectal malformations, Genitourinary anomalies, Vesicoureteric reflux

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### INTRODUCTION

Anorectal malformations (ARM) are diagnosed during post-natal examination when there is absence or ectopic position of anus. It is a complex group of disorders and usually associated with anomalies of other systems too.<sup>1</sup> Anorectal malformation can be high or low, which depends upon the level at which rectum ends in relation to pubo-coccegeous line. There is variability in data however about 1 in 5000 live births has anorectal malformation.

Genitourinary anomalies are commonly associated with ARM which has been recently reviewed. The reported incidence of urinary tract abnormalities in these patients range from 26% to 50% in several studies.<sup>2,3</sup> Urological anomalies need extensive investigations however most of the abnormalities of genitalia are diagnosed on careful clinical examination. Anorectal malformations and their association with abnormalities of genitourinary tract have been reviewed lately. The incidence of genitourinary anomalies varies with level of ARM<sup>4</sup>, i.e. the higher

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the level of rectum, increased incidence of genitourinary anomalies.<sup>5-7</sup> Genitourinary anomalies were seen in 49.6% patients with intermediate or high-level anorectal lesions. Different genitourinary anomalies have been associated with ARM which includes vesicoureteric reflux (VUR), ureteropelvic junction obstruction (UPJO), hydronephrosis, hypospadias, renal agenesis and undescended testis (UDT).<sup>8</sup> 52.9% were shown to have VUR.<sup>4</sup> In Netherlands 50% of patients with anorectal malformation were having genitourinary abnormalities<sup>9</sup> while in Romania the reported incidence was 24%.<sup>10</sup> A study conducted in Bangladesh showed the incidence of urinary tract abnormalities relatively higher to 71% among patients with anorectal abnormalities.<sup>11</sup>

The aim of our study is to evaluate the frequency of coexisting genitourinary tract anomalies in children with anorectal malformations, which can be a source of significant morbidity. The difference in reports on the frequency of associated anomalies with anorectal malformation requires further studies.

## METHODOLOGY

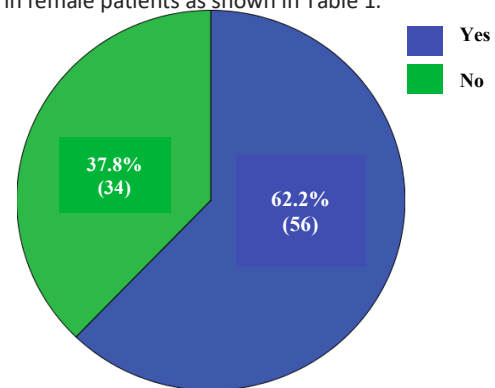
This descriptive cross sectional study was carried out in Department of Pediatric Surgery, Khyber Teaching Hospital, Peshawar from August 2017 To February 2018. Ethical approval was obtained from Institutional ethical review board. Any child (both genders) with clinically apparent diagnosis of anorectal malformation, age 1 month or less (Neonate) was included in the study. Patients who underwent any surgical intervention elsewhere for anorectal malformation and cases of intersex or genital ambiguity were excluded. The sample size was 90 and sampling technique non-probability consecutive sampling. All patients meeting the inclusion criteria admitted in the ward were included in the study. The purpose and benefits of the study were explained to the child guardian and written informed consent was obtained. The patient's sex, anorectal lesion level and the presence of urinary tract and genital anomalies were recorded. Level of anorectal lesion was determined by radiographic evaluation. Renal ultrasound and intravenous-pyelography (IVP) and Voiding Cysto-urethrogram (VCUG) were performed for urinary tract malformations in all cases with high level anorectal lesions. In low level anorectal lesion ultrasound was performed initially with IVP and VCUG performed later if needed to investigate any anomaly (dilated ureters, hydronephrosis) found on ultrasound. All the relevant data were recorded in a pre-designed printed proforma.

Confounders and bias in the study were controlled by strictly following the inclusion and exclusion criteria. SPSS 20.0 was used for analysis of data. Mean  $\pm$  standard deviation was calculated for quantitative variables such as age. Percentage and proportion were calculated for categorical variables such as gender, genitourinary anomalies and lesion level. Genitourinary anomalies were stratified among age, gender and lesion level (high or low), genital ambiguity, previous surgery elsewhere to see effect

modification. Post stratification was done through Chi square test keeping P value <0.05 was considered significant. All results were presented in the form of table and graphs.

## RESULTS

A total of 90 patients presenting with anorectal malformations were included in the study. There were 62(68.89%) males and 28(31.11%) females. Male to female ratio was 2.2:1. Average age of the patients was  $5.03 \pm 6.11$  days with range of 1 day to 29 days. The frequency of genitourinary abnormalities among children with anorectal malformations is shown in Figure 1. Gender wise distribution shows that although ARM is more common in male patients, the frequency of associated genitourinary anomalies is more in female patients as shown in Table 1.



**Figure 1** frequency of genitourinary abnormalities among children with anorectal malformations (n=90)

		Genitourinary Abnormalities		p-value
		Yes	No	
Gender	Male	35 56.5%	27 43.5%	0.093
	Female	21 75.0%	7 25.0%	

**Table 1** gender wise distribution of genitourinary abnormalities in anorectal malformations.

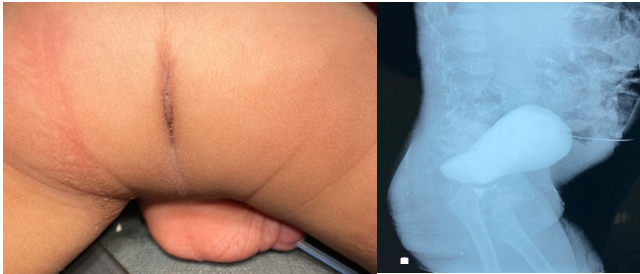
The common types of anomalies found are shown in Table 2. Similarly it is also found that genitourinary abnormalities are more common in patients with High ARM. Table 3. Figure 2a and b.

Type of Anomaly	Frequency	Percent
Vesicoureteric reflux (VUR)	21	23.3
Hypospadias	13	14.4
Ureteropelvic junction obstruction (UPJO)	8	8.9
Renal agenesis	3	3.3
Undescended testis (UDT)	6	6.7
Renal ectopia	5	5.6
Total Anomalies	56	62.2
No Anomalies	34	37.8
Total Patients	90	100.0

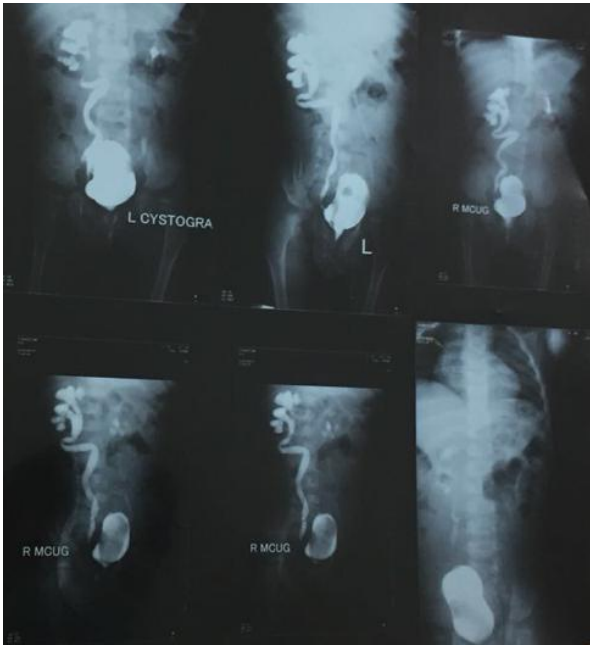
**Table 2** showing common types of anomalies (n=90)

		type of anomaly						Total
		VUR	Hypospadias	UPJO	renal agenesis	UDT	renal ectopia	
		Count	Count	Count	Count	Count	Count	
level of ARM	Low	3	9	0	2	0	5	19
	High	18	4	8	1	6	0	37

**Table 3** genitourinary abnormalities are more common in patients with High ARM



**Fig 2 a** ARM with no fistula (High ARM), **Fig b**, Colostogram of same patient showing (High ARM).



**Figure 3** VCUG showing right sided VUR

## DISCUSSION

Patients with Anorectal Malformations (ARM) have co-existing anomalies of one or more other systems as well. These include cardiac, vertebral, gastrointestinal etc however genito-urinary system abnormalities are most commonly associated.<sup>12</sup>

As stated earlier, ARM is complex congenital abnormality that ranges from simple anal stenosis to anterior transposition of anus to extremely complex anomalies involving distal portion of hindgut and urogenital organs.<sup>13</sup> The reported incidence is 1 in 2500 to 5000

live births with slight male predominance<sup>14,15</sup> as same was the case in our study.

About 26-70% of ARM patients have genitourinary anomalies.<sup>1,2</sup> In this study it was 62.2%. Most commonly found urinary anomaly in this study was Vesicoureteric reflux (VUR) (Figure 3). Various studies have reported Vesicoureteric Reflux from 19 to 47.2% in patients with ARM<sup>5,16</sup> while in the present study it was observed to be 21(23%). Metts and Boemers found VUR in 32% of their cases.<sup>17</sup> Misra *et al.* reported VUR in 7.5% of patients with low deformity<sup>18</sup> but Tohda and Moore reported the incidence of VUR only in 0.7% and 5.4% of their patients.<sup>19</sup> The difference in results is due to variability in the study methods and criteria for investigating the genitourinary anomalies. In some studies VCUg was done only after abnormal ultrasound findings.<sup>12</sup>

Other urological abnormalities found were Uretero-pelvic junction obstruction (UPJO), renal ectopia and renal agenesis. Similar abnormalities are also reported in literature.<sup>8,9</sup> Among genital abnormalities most common anomaly found was hypospadias 13(14.4%) followed by undescended testis 6(6.6%). Hoekstrn *et al.*<sup>20</sup> and Vaishali reported incidence of hypospadias in their studies as 6% and 4.55%, respectively. Slightly increased incidence of hypospadias in our study needs evaluation for other factors.

Increased incidence of genitourinary anomalies was found in this study with high ARM (41.11%) as compared to Low ARM (21.11%). McLorie *et al.* reported figures of 20% and 60% respectively.<sup>5</sup> Parrot found incidence of 14% with low and 40% with high anomalies.<sup>21</sup> Fourteen percent of patients had genital anomalies and 25.6% had urologic anomalies in a study conducted by Sangkhathat *et al.*<sup>5</sup> while in our study urologic anomalies were 41% and overall genital abnormalities were 21.11%.

The most common abnormality in our study associated with low ARM was hypospadias, seen in 9 cases (10%) while that of high ARM was VUR (20%). The reported incidence of hypospadias by Hoekstra *et al.* is 6%.<sup>22</sup> Although renal agenesis has been the most commonly reported anomaly in literature, we have encountered only 2 cases (2.2%) in our study. Surprisingly renal ectopia was the most commonly seen anomaly (5.6%) while the incidence of VUR was 3.33% with low ARM as against 47% by Rich *et al.*<sup>23</sup> and 8.6% by Sangkhathat *et al.*<sup>5</sup>

It is shown by our study that the incidence and type of genitourinary anomalies varies with level of anorectal malformation. Goossens *et al.* observed when the defect is lower down, the incidence of genitourinary anomalies are less. While incidence increases in higher level defects such as rectovesical fistula, the higher is incidence of

genitourinary anomalies are raised.<sup>9</sup> The findings of our study cannot be generalized as it is a single centre study further studies with large cohort and multiple centres are required to assess the amplitude to these anomalies in the country.

## CONCLUSION

ARM patients should be screened for urogenital anomalies. Early detection of the associated anomalies will result in proper management and better outcome. Also awareness about associated anomalies with ARM, among general population as well as paediatricians is necessary for early referral to pediatric surgeon.

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