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Original Research Article

Frequency of Genitourinary Abnormalities in Patients Having Anorectal Anomalies

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Abstract: *Anorectal malformations (ARMs) are frequently associated with genitourinary anomalies, which can lead to significant morbidity if left undiagnosed. Early detection is critical for optimal clinical management and prognosis.* ***Objective:*** *To determine the frequency and pattern of genitourinary abnormalities in neonates diagnosed with anorectal malformations.* ***Methods:*** *A descriptive cross-sectional study was conducted at the Department of Pediatric Surgery, Lady Reading Hospital, Peshawar, Pakistan, from 05-07-2024 to 05-01-2025. One hundred fifty-one neonates aged 1–30 days with radiologically confirmed ARMs were enrolled through non-probability consecutive sampling. Neonates with prior surgical interventions or genital ambiguity were excluded. Data on demographic variables and ARM type (high vs. low lesion) were collected. Genitourinary anomalies were identified through clinical examination, ultrasound, voiding cystourethrogram (VCUG), and intravenous pyelography. Statistical analysis was performed using SPSS version 25, and p-values <0.05 were considered significant.* ***Results:*** *Out of 151 neonates, 89 (58.9%) were male and 62 (41.1%) were female. Genitourinary anomalies were present in a substantial proportion of patients. The most common abnormality was vesicoureteral reflux (VUR), found in 52 cases (34.4%), followed by hypospadias in 19 cases (12.6%), ureteropelvic junction obstruction (UPJO) in 18 cases (11.9%), and undescended testis (UDT) in 8 cases (5.3%). Hypospadias and UDT were exclusively observed in male neonates ($p < 0.001$). High-type ARMs (n=63, 41.7%) were significantly associated with increased genitourinary anomaly frequency.* ***Conclusion:*** *Genitourinary anomalies, particularly vesicoureteral reflux, are common in neonates with anorectal malformations, especially those with high-type lesions. Routine screening with ultrasound and VCUG is essential to identify and manage these associated abnormalities early, thereby improving long-term outcomes.*

Keywords: Anorectal malformations, genitourinary abnormalities, vesicoureteral reflux, hypospadias, undescended testis

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Introduction

Anorectal malformations (ARM) are recognized in post-natal examination as there is lack or ectopic location of anus. It is a complicated collection of illnesses usually linked with anomalies of various systems (1). Anorectal malformation may occur high or low, depending upon the level where the rectum stops in relation to the pubo-coccyx line. There is fluctuation in data, yet roughly 1 in every 5000 live newborns has anorectal abnormality. Genitourinary abnormalities are usually related to ARM, which has been recently examined. In numerous investigations, the estimated prevalence of urinary tract anomalies in these patients varied from 26% to 50% (2, 3). While the precise cause of ARM is unknown, hereditary factors likely play a role in its development. The chance of having a second kid with an anorectal abnormality is around one percent (4). There are various genetic syndromes with an elevated frequency of ARM, like the Currarino triad, which shows the inheritance of autosomal dominant genes, and people with trisomy 21 have a recognized link with anorectal deformity without fistula (5).

Urological anomalies need detailed studies, yet most of the anomalies of the genitalia are recognized by careful clinical examination. Anorectal anomalies and their connection with defects of the genitourinary tract are being examined lately. The occurrence of genitourinary anomalies rises with the level of ARM (6). Different genitourinary defects have been connected with ARM such as vesicoureteric reflux, ureteropelvic junction blockage, hydronephrosis, hypospadias, renal agenesis as well as undescended testis (7) Genitourinary abnormalities like vesicoureteric reflux 23.3%, ureteropelvic junction obstruction 8.9%, hypospadias 14.4%, and undescended testis 6.7% was observed in patients with anorectal anomalies (7). In a study, 50% of patients with anorectal malformation experienced genitourinary abnormalities (8). whereas in a study, the frequency was 24% (9). Another study demonstrated that the incidence of urinary tract abnormalities was considerably greater, at 71%, amongst those with ARM (10).

The goal of this study is to investigate the frequency of concurrent urinary tract Malformations in patients with imperforate anus. Genitourinary abnormalities among individuals with Imperforate anus are a source of substantial morbidity. Variability in reports on the frequency of related abnormalities with anorectal malformation demands further studies on this subject.

Methodology

The study was conducted as a descriptive cross-sectional investigation within the Department of Pediatric Surgery at Lady Reading Hospital, Peshawar, from 05-07-2024 to 05-01-2025. A sample size of 151 participants was determined using statistical calculations accounting for a 95% confidence interval, an anticipated prevalence of 6.7% for undescended testis[7] as a representative genitourinary anomaly, and a precision margin of 4%. Participants were recruited through consecutive sampling, prioritizing accessibility and clinical presentation over probabilistic methods.

Eligibility criteria focused on neonates aged 1 to 30 days diagnosed with anorectal malformations, irrespective of gender. Cases involving prior surgical interventions for the malformation or instances of genital ambiguity were excluded to minimize confounding variables. Ethical clearance was secured from the institutional review board, and informed consent was obtained from guardians after explaining the study's objectives, procedures, and voluntary nature of participation. Data collection involved systematic recording of demographic details such as age, gender, and anthropometric measurements, followed by clinical evaluations for genitourinary abnormalities, including vesicoureteral reflux, ureteropelvic junction obstruction, hypospadias, and undescended

testis. These assessments were conducted under the supervision of a consultant with over five years of post-fellowship expertise, ensuring consistency and accuracy.

Clinical parameters, including lesion level (high or low) and the presence of genital ambiguity, were documented using a structured proforma. Diagnostic imaging and physical examinations were employed to identify abnormalities, adhering to predefined operational definitions. For data analysis, SPSS version 23 was utilized, with numerical variables such as age and BMI expressed as mean ± standard deviation or median and interquartile range, depending on distribution normality assessed via the Shapiro-Wilk test. Categorical variables, including gender and anomaly frequencies, were summarized as percentages. Stratification by variables such as lesion level and gender was performed to explore associations, with statistical significance evaluated using chi-square or Fisher's exact tests at a 5% threshold.

Results

The study included a cohort of 151 patients with a mean age of 16.41 ±8.96 days and an average BMI of 13.66 ±0.93 kg/m². Males constituted the majority of participants (89 cases, 58.9%), while females accounted for 62 cases (41.1%). Lesion level showed higher lesions observed in 88 patients (58.3%) and lower lesions in 63 patients (41.7%). Genital ambiguity was rare in only 2 cases (1.3%), and the majority (144 cases, 95.4%) had no prior surgical interventions.

Genitourinary abnormalities were prevalent, with vesicoureteral reflux (VUR) identified as the most common anomaly, affecting 52 patients (34.4%). Ureteropelvic junction obstruction (UPJO) was observed in 18 cases (11.9%), while hypospadias and undescended testis (UDT) were present in 19 (12.6%) and 8 (5.3%) patients, respectively. Stratification by gender revealed distinct patterns: VUR showed no significant disparity, occurring in 30 males (57.7% of VUR cases) and 22 females (42.3%). Similarly, UPJO was marginally more frequent in males (12 cases, 66.7%) than in females (6 cases, 33.3%), though this difference was not statistically notable.

Hypospadias and UDT were exclusively male-specific anomalies, with all 19 hypospadias cases and 8 UDT cases occurring in males ($p<0.001$ and $p = 0.01$, respectively). No notable association of abnormalities with age, BMI, previous surgical intervention, lesion level, and genital ambiguity was observed ($p > 0.05$).

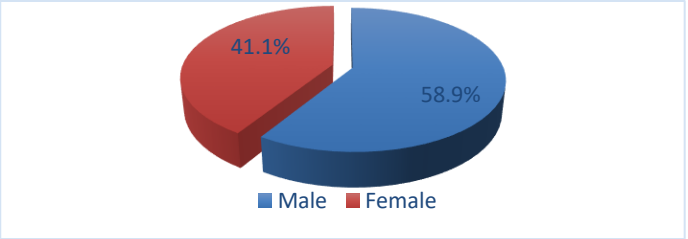

Figure 1: Gender distribution.

Table 1: Clinical presentation

Clinical presentation		N	%
Lesion level	High	88	58.3%
	Low	63	41.7%
Genital ambiguity	Yes	2	1.3%
	No	149	98.7%
Previous surgical intervention	Yes	7	4.6%
	No	144	95.4%

Table 2: Frequency of genitourinary abnormalities

Genitourinary abnormalities		N	%
Vesicoureteral Reflux (VUR)	Yes	52	34.4%
	No	99	65.6%
Ureteropelvic Junction Obstruction (UPJO)	Yes	18	11.9%
	No	133	88.1%
Hypospadias	Yes	19	12.6%
	No	132	87.4%
Undescended Testis (UDT)	Yes	8	5.3%
	No	143	94.7%

Table 3: Stratification of genitourinary abnormalities by gender

Genitourinary abnormalities		Gender				P value
		Male		Female		
		N	%	N	%	
Vesicoureteral Reflux (VUR)	Yes	30	57.7%	22	42.3%	0.82
	No	59	59.6%	40	40.4%	
Ureteropelvic Junction Obstruction (UPJO)	Yes	12	66.7%	6	33.3%	0.47
	No	77	57.9%	56	42.1%	
Hypospadias	Yes	19	100.0%	0	0.0%	0.0001
	No	70	53.0%	62	47.0%	
Undescended Testis (UDT)	Yes	8	100.0%	0	0.0%	0.01
	No	81	56.6%	62	43.4%	

Discussion

The study presented data on a cohort of 151 patients with anorectal malformations (ARM), with a mean age of 16.41 ± 8.96 days and an average BMI of 13.66 ± 0.93 kg/m². This cohort primarily consisted of male patients, with 89 male participants (58.9%) and 62 female participants (41.1%), mirroring the well-documented trend that males are more commonly affected by anorectal malformations than females. In line with expectations, most patients (58.3%) had high lesions, while 41.7% presented with low lesions. This distribution highlights the varied nature

of ARM, with high lesions often associated with more severe anatomical and functional implications. Similar distributions have been observed in several studies, where most ARM cases presented with high lesions (5).

The finding of genital ambiguity in only 1.3% of the cases is noteworthy, as this condition is generally considered an important marker for more complex ARM cases, particularly those with associated chromosomal or hormonal abnormalities. Genital ambiguity in ARM patients is less common in non-syndromic cases, as noted in our study. Previous research has reported a higher incidence of genital ambiguity, particularly in patients with associated syndromes like VACTERL or CAKUT (congenital anomalies of the kidney and urinary tract) (11). Our findings suggest that genital ambiguity is not common in the general ARM population. However, assessing it in patients with more complex or syndromic forms of ARM is critical. This observation is consistent with the findings of Levitt et al., who suggested that genital ambiguity is typically seen in cases with more complex ARM and syndromic associations (5).

The study also examined the occurrence of genitourinary abnormalities, with vesicoureteral reflux (VUR) emerging as the most common anomaly, affecting 34.4% of the cohort. The high prevalence of VUR in ARM patients aligns with findings from previous studies, where VUR is consistently identified as one of the most common genitourinary anomalies in this population (11,12). Interestingly, the study found no notable gender differences in the prevalence of VUR, with 57.7% of cases occurring in males and 42.3% in females. This lack of a significant gender disparity is in contrast to some studies, which have reported a higher incidence of VUR in males, possibly due to differences in urinary tract anatomy or the presence of other urological malformations (12).

Another noteworthy finding was the prevalence of ureteropelvic junction obstruction (UPJO), which occurred in 11.9% of the patients. This rate is consistent with the findings of a study, where UPJO was commonly seen in ARM patients, though the exact prevalence can vary depending on the cohort's complexity (11). While not statistically notable, the marginally higher incidence of UPJO in males (66.7%) in our study is consistent with findings in the literature suggesting a male preponderance in certain urological anomalies (13). However, it is important to note that our study's difference between male and female patients was insufficient to draw definitive conclusions regarding a gender-specific risk for UPJO in ARM cases.

The study also found that hypospadias and undescended testis (UDT) were exclusively male-specific anomalies, with hypospadias affecting 12.6% of male patients and UDT affecting 5.3%. Both of these anomalies have been consistently linked to anorectal malformations in male patients in previous research, with hypospadias being one of the most common genital abnormalities associated with ARM (11). The gender specificity of these anomalies in our study highlights the importance of conducting gender-specific evaluations in clinical practice, particularly for male patients with ARM, to ensure that conditions like hypospadias and UDT are detected and managed appropriately. The strong association between male gender and genital abnormalities in the context of ARM is well-documented in the literature. It underscores the necessity for careful and timely assessments for males presenting with anorectal malformations (5).

One of the significant aspects of our study is the low incidence of previous surgical interventions, with only 4.6% of patients having undergone prior surgical procedures. This is relatively low compared to a study where previous surgeries, such as colostomies or initial corrective operations, are more common in ARM patients (5). Our study's rarity of prior surgical interventions suggests that many patients in this cohort were either newly diagnosed or had not yet required surgical management. The high proportion of patients without prior surgeries indicates that these infants were likely in the early stages of diagnosis or management, and their outcomes may depend heavily on the timing of surgical intervention and follow-up care.

The findings of this study are consistent with many established patterns in the literature regarding the association between anorectal malformations and genitourinary anomalies. VUR and UPJO were commonly observed, with hypospadias and UDT being strongly associated with male gender, reflecting known trends in the pathophysiology of ARM and its associated conditions. The study emphasizes the need for comprehensive screening, including urological and genital assessments, for all ARM patients, particularly males, to ensure that associated abnormalities are detected early and managed appropriately. It is also critical to recognize the rare occurrence of genital ambiguity in non-syndromic cases, which underscores the need for careful evaluation in complex or syndromic cases of ARM.

Conclusion

Genitourinary abnormalities were prevalent in anorectal malformations, with vesicoureteral reflux (34.4%), hypospadias (12.6%), ureteropelvic junction obstruction (11.9%), and undescended testis (5.3%) observed. Routine screening via imaging and clinical exams, particularly for males and high-risk lesions, is recommended to mitigate morbidity and guide timely interventions.

Declarations

Data Availability statement
All data generated or analysed during the study are included in the manuscript.

Ethics approval and consent to participate
Approved by the department concerned. (IRB-REF-37/LRH/MTI)

Consent for publication
Approved

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Conflict of interest

The authors declared the absence of a conflict of interest.

Author Contribution

IUD (Postgraduate Resident),
Manuscript drafting, Data acquisition, Study Design, Data entry, and Data analysis.

MYK (Associate Professor)
Review of Literature, Conception of Study, Development of Research Methodology Design,

All authors reviewed the results and approved the final version of the manuscript. They are also accountable for the integrity of the study.

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