

FREQUENCY OF UROGENITAL ANOMALIES IN PATIENTS PRESENTING WITH HYPOSPADIAS IN A TERTIARY CARE HOSPITAL

Romila Mazhar^{1*}, Muhammad Anwar¹, Amna Choudry¹

¹Paediatric Surgery, National Institute of Child Health

*Corresponding author: Romila Mazhar, Email: romilamazhar@gmail.com

ABSTRACT

Objective: To determine the frequency of urogenital anomalies in patients presenting with hypospadias in a tertiary care hospital.

Study Design: Descriptive Cross-sectional

Place and Duration of the Study: Department of Paediatric Surgery, NICH from 13th May 2026 to 13th August 2026.

Methodology: The patients were 196 male patients aged 3-10 years with clinically diagnosed hypospadias who were consecutively selected. A clinical and ultrasonographic assessment of the urogenital anomalies was conducted, such as inguinal hernia, undescended testes condition, ambiguous genitalia and renal anomalies. SPSS-26 was used to analyse the data.

Results: The mean age was 6.92 ± 3.17 years. The most common anomaly was undescended testes, 45 (22.9%); followed by inguinal hernia and renal anomalies, 15 (7.7%) each; and ambiguous genitalia, 12 (6.1%). There was a greater incidence of urogenital anomalies in children between 3 and 7 years of age than in children >7 years of age (67.1% vs 32.9, $p = 0.092$). There was a significant association between the anatomical type of hypospadias and the occurrence of a urogenital anomaly ($p < 0.001$).

Conclusion: Urogenital anomalies were commonly found in patients who presented with hypospadias, with the most common being undescended testes, followed by inguinal hernias, renal anomalies, and ambiguous genitalia. Younger children (3-7 years old) showed a relatively higher percentage of urogenital anomalies, but there was an insignificant association between age group and the presence of urogenital anomalies. A while highly significant association was noted between the hypospadias type and the presence of urogenital anomalies.

KEYWORDS: Hypospadias; Urogenital Abnormalities; Cryptorchidism; Hernia, Inguinal; Child, Preschool; Tertiary Care Centers; Pakistan

INTRODUCTION

Hypospadias is one of the most important congenital malformations of the male genitourinary system that affects about 1 in every 200 to 300 live male neonates [1]. A population-based, registry-based estimate of the global burden of the defect places it at 83.0 cases per 10,000 live male births, making it the second most common urogenital birth defect in boys after cryptorchidism [2]. It occurs embryologically when there is incomplete fusion of urethral folds on the underside of the penile shaft, with the external urethral meatus in an abnormal position, which results in the characteristic triad of foreskin abnormality, penile shaft curvature, and a wide penile head (glans tip) [3]. Regarding anatomical distribution, 70% of affected males have a distally located meatus from the glans to the penile shaft, while the remaining 30% have variants that are located more proximally, on the glans penoscrotal, scrotal and even perineal, with significantly more complex reconstructive options.

It is known that hypospadias is a multifactorial disorder and that both genetic predisposition and in-utero exposure to environmental factors during a critical period in urethral development are likely to play a role in its aetiology [4]. The genetic component is also clinically important: If a father suffers from hypospadias, a quarter of his sons are likely to suffer from the same defect, suggesting it is polygenically inherited [4]. Mechanistically, it has been shown that disruption of androgen-dependent signalling during urethral tubularisation interferes with the normal migration of the meatal opening to the glans apex, and compensatory overgrowth of the dorsal preputial skin causes the ventral curvature that is characteristically referred to as 'chordee' [5].

The primary penile malformation is frequently not the sole anomaly encountered at presentation. Depending on the series examined, concurrent congenital defects are documented in 7% to 40% of hypospadias cases [6]. Urogenital malformations constitute the most prevalent anomaly category, accounting for 40% of all anomalies co-identified with hypospadias, and comprising conditions such as cryptorchidism, inguinal hernia, vesicoureteral reflux, ureteropelvic junction obstruction, and bladder exstrophy [6]. Structural renal pathology, including unilateral renal agenesis and

dysgenesis kidneys, is similarly over-represented, a finding concordant with the shared mesonephric ductal origin of the urinary and reproductive tracts [7]. Despite this recognised association, granular local data on the prevalence distribution of specific urogenital anomalies within hypospadias cohorts remain limited.

There is a wide range of published prevalence rates for each setting. In a Pakistani referral series, Ahmad et al, found that inguinal hernia was the most common co-occurring anomaly (37%), cryptorchidism (30%), and ambiguous genitalia (15%) [8]. However, a review of the hospital population in Indonesia by Hariyono et al. reported a much lower prevalence of cryptorchidism, 10.7% [9], and a review of Patra et al. in the eastern part of India reported 8.27% cryptorchidism and 6.89% inguinal hernia [10]. These inter-institutional differences underscore how referral patterns, surgical case-mix, and diagnostic protocols can substantially shape the anomaly spectrum observed and reinforce the value of locally generated prevalence data for clinical planning [8-10].

From a surgical standpoint, the concurrent presence of urogenital anomalies has direct implications for operative management, as co-existing defects often necessitate additional or staged interventional procedures to achieve satisfactory functional and aesthetic outcomes [11]. Comprehensive systematic reviews have delineated the respective contributions of genetic vulnerability and endocrine-disrupting environmental exposures to the pathogenesis of hypospadias [12], while recent experimental work has clarified the embryonic cellular mechanisms governing urethral fold fusion [13]. Maternal exposure to progestogenic agents during organogenesis has been proposed as a hormonal risk modifier [14]. Recent evidence also suggests that environmental endocrine disruptors may contribute to abnormalities in male reproductive development, including hypospadias and associated urogenital anomalies [15]. Low birth weight has independently emerged as an associated risk factor [16], and large-scale population-based surveillance has further characterised severity-stratified prevalence trends alongside demographic risk distributions [17].

An estimated 20.9 per 10,000 live male births are associated with hypospadias and other congenital anomalies, most of which are urogenital, which are a significant burden on paediatric surgical services, and a meaningful contributor to patient morbidity. However, there is limited epidemiological data in the Pakistani context, and it is locally sourced. The present study was therefore performed to establish the prevalence of urogenital anomalies in a tertiary paediatric urological referral centre in male children with hypospadias and to help surgical planning and to improve long-term patient outcomes, to have a systematic description of the co-occurring anomalies.

METHODOLOGY

The study was a descriptive cross-sectional study which was carried out at the Department of Paediatric Surgery, National Institute of Child Health, Karachi, from the ERC and CPSP. Informed written consent from each child's parent/carer was obtained before their enrolment. Clinically confirmed hypospadias children (males) aged 3–10 years were selected using consecutive sampling (non-probability sampling). The diagnosis rested on physical examination revealing an abnormally located external urethral meatus on the ventral penile surface; each case was then sub-typed as glanular, coronal, sub-coronal, distal penile, mid-penile, proximal penile, penoscrotal, or perineal based on the position of the meatus, for stratified analysis, hypospadias subtypes were grouped a priori into distal (glanular, coronal, sub-coronal, distal penile), mid (mid penile), and proximal (proximal penile, penoscrotal, scrotal) categories. Children referred from other centres specifically for hypospadias repair, those with a documented history of prior failed corrective surgery, and those attending for a staged hypospadias procedure were excluded from participation. Four urogenital anomalies were assessed using pre-specified clinical criteria. Inguinal hernia was recorded when an inguinoscrotal swelling was identified while the child stood upright, and at least one additional sign was present; a cough impulse felt on palpation, failure to reach the upper limit of the swelling on examination, or a swelling that reduced fully, partially, or not at all with manual pressure. Undescended testes were noted when one or both testes were absent from the scrotal sac; in such cases the ipsilateral inguinal canal was palpated, and where the testis remained impalpable, ultrasonography was requested to locate it within the pelvis or abdomen. Ambiguous genitalia were diagnosed when any two of the following five signs coexisted: a single or paired gonad (testicular or ovarian in character), a phallus of intermediate proportions between a penis and a clitoris, ventral penile curvature (chordee), a small or bifid scrotum with poorly developed genital folds, or labioscrotal folds that were either fused or separated. Renal anomalies comprised two conditions: renal agenesis, identified ultrasonographically as complete absence of a kidney, and renal anomalies comprised two conditions: renal agenesis, identified ultrasonographically as complete absence of a kidney, and renal dysgenesis, defined as sonographic evidence of abnormal renal development, operationalised as a renal length below the third percentile or above the ninety-seventh percentile of age-matched paediatric reference values (Rosenbaum nomogram), an ectopic renal position outside the renal fossa, or cortical thickness below 8 mm in children younger than 6 years and below 11 mm in older children. The sample size of 196 was derived using the W.H.O calculator, frequency of ambiguous genitalia in hypospadias patients (15%)⁸, 5% of precision and a 95% C.I. After confirming eligibility, the principal investigator recorded each child's demographic details and relevant clinical findings on a structured proforma. All participants underwent full physical examination together with abdominal ultrasonography to detect any co-existing urogenital anomalies, and observations were

entered into the proforma on the same occasion. All ultrasound examinations were performed by consultant radiologists experienced in paediatric ultrasonography using standard paediatric abdominal ultrasonography equipment. The radiologists were not involved in the clinical classification of hypospadias at the time of image interpretation. Data were entered and analysed in SPSS-26; frequency with percentage was computed for urogenital anomalies and hypospadias subtypes, mean $\pm$ SD for age, and Chi-square test applied post-stratification ($p \leq 0.05$).

RESULTS

The baseline characteristics of the 196 study participants are presented in **Table I**. The mean age of the cohort was 6.92 ± 3.17 years (range 3–10; 95% CI 6.48–7.37). Most of the participants came from the urban areas (151, 77.0%) and 45 (23.0%) from the rural areas. The most common type of hypospadias was the penoscrotal type (52, 26.5%), then coronal (33, 16.8%), then sub-coronal (32, 16.3%), and lastly glanular (28, 14.3%). Distal penile and mid-penile hypospadias were reported in 19 (9.7%) each, and proximal penile 8 (4.1%) and scrotal 5 (2.6%) were relatively uncommon. Concerning the urogenital anomalies, 15 patients were found with inguinal hernia and renal anomalies, respectively (7.7%). Forty-five cases of undescended testes were classified as abdominal testicles (6.6%) and inguinal testicles (16.3%). The cases of ambiguous genitalia were 12 (6.1%).

Table II presents the association of urogenital anomalies with age group and type of hypospadias among the 196 patients. Urogenital anomalies were observed in 51 (67.1%) patients aged 3–7 years and in 25 (32.9%) patients older than 7 years. Among those without urogenital anomalies, 66 (55.0%) were in the 3–7 years age group and 54 (45.0%) were older than 7 years. The association between age group and the presence of urogenital anomalies was not statistically significant ($p=0.092$). A statistically significant association was observed between the type of hypospadias and the presence of urogenital anomalies ($p<0.001$). Among patients with urogenital anomalies, the most common type of hypospadias was penoscrotal (40.8%), followed by coronal (15.8%), glanular (9.2%), distal penile (9.2%), mid penile (9.2%), sub-coronal (7.9%), scrotal (6.6%), and proximal penile (1.3%). In contrast, among patients without urogenital anomalies, the most frequent types were sub-coronal (21.7%), glanular (17.5%), coronal (17.5%), penoscrotal (17.5%), distal penile (10.0%), mid penile (10.0%), and proximal penile (5.8%), while no scrotal hypospadias cases were observed in this group. These findings demonstrate a significant association between the anatomical type of hypospadias and the presence of urogenital anomalies.

Table I: Baseline Characteristics of Study Participants (n=196)			
Mean ± Standard Deviation			95% Confidence Interval
Age in years = 6.92 ± 3.17			6.48–7.37
Frequency (%)			
Residential Status	Urban		151 (77.0)
	Rural		45 (23.0)
Type of Hypospadias	Glanular		28 (14.3)
	Coronal		33 (16.8)
	Sub-coronal		32 (16.3)
	Distal Penile		19 (9.7)
	Mid Penile		19 (9.7)
	Proximal Penile		8 (4.1)
	Penoscrotal		52 (26.5)
	Scrotal		5 (2.6)
Urogenital Anomalies	Inguinal Hernia		15 (7.7)
	Undescended Testes	Abdominal	13 (6.6)
		Inguinal	32 (16.3)
	Ambiguous Genitalia		12 (6.1)
	Renal Anomalies		15 (7.7)

Table II: Association of Urogenital Anomalies with Age Group and Type of Hypospadias among Patients with Hypospadias (n=196)			
Age Group	Urogenital Anomalies		P-Value
	Yes	No	
3–7	51 (67.1)	66 (55.0)	0.092
>7	25 (32.9)	54 (45.0)	
Type of Hypospadias	Urogenital Anomalies		P-Value
	Yes	No	
Glanular	7 (9.2)	21 (17.5)	<0.001
Coronal	12 (15.8)	21 (17.5)	
Sub-coronal	6 (7.9)	26 (21.7)	
Distal Penile	7 (9.2)	12 (10.0)	
Mid Penile	7 (9.2)	12 (10.0)	
Proximal Penile	1 (1.3)	7 (5.8)	
Penoscrotal	31 (40.8)	21 (17.5)	
Scrotal	5 (6.6)	0 (0.0)	

DISCUSSION

In addition to anatomical variations, various other developmental anomalies are associated with hypospadias, which may make surgical correction more challenging and cause a decrease in functional performance with correction [1,6,11], making it a clinically important congenital defect. The most common defect is ventrally displaced urethral meatus, and associated urogenital defects are very common and should be fully evaluated when the animal is presented. However, there is still significant epidemiological variability and related congenital burden because of surveillance data that do not rely on the population, and this justifies the need for more organised surveillance measures [2,20].

Of these 196 children with a mean age of 6.92 ± 3.17 years, the most common hypospadias subtypes were penoscrotal (26.5%), coronal (16.8%), and sub-coronal (16.3%). Overall, there were many urogenital anomalies. Undescended testes in 45 (22.9%) patients, 6.6% abdominal testes and 16.3% inguinal testes were detected as the most common abnormalities. In 7.7%, an inguinal hernia was detected, and in 7.7% a renal anomaly was detected, and in 6.1% ambiguous genitalia were detected.

Compared to Ahmad et al. [8], who reported inguinal hernia (37%), undescended testes (30%), and ambiguous genitalia (15%), the current study has significantly lower rates of inguinal hernia (7.7% vs. 37%), undescended testes (22.9% vs. 30%), and ambiguous genitalia (6.1% vs. 15%).

Tariq et al. reported cryptorchidism in 18.4% and inguinal hernia in 12.1% of hypospadias patients managed at a tertiary military hospital [21], while Hussain et al. documented undescended testes in 19.7% of cases from a Lahore-based cohort [22]. These findings are comparable with the present study, where cryptorchidism was identified in 22.9% of participants. Such differences could be due to variations in case mix, referral patterns, and distribution of disease severity. In the present study, a statistically significant association was also observed between the type of hypospadias and the presence of urogenital anomalies ($p < 0.001$), with proximal variants particularly penoscrotal and scrotal types showing a higher frequency of associated anomalies. This observation is consistent with previous reports indicating that more severe or proximal forms of hypospadias are more frequently accompanied by additional urogenital defects. In contrast, Hariyono et al. (9) documented cryptorchidism (undescended testes) in 10.7% of their cohort, which is approximately half the frequency observed in the present study (22.9%). Similarly, Patra et al. [10] reported undescended testes in 8.27% and inguinal hernia in 6.89% of patients, figures closer to the current 7.7% for inguinal hernia but markedly lower for undescended testes. Our cohort showed a higher cryptorchidism rate, and this might reflect a higher rate of proximal or severe hypospadias, which is known to be associated with testicular maldescent.

On the embryological level, hypospadias and undescended testes are similarly developed with a common pathway with androgen-dependent differentiation of the genital tubercles and migration of gubernacular cell types [3,5]. Therefore, change in androgen signalling and/or gene-environment interactions may be evident when they occur concurrently [12,15]. Experimental studies have also helped to elucidate the cellular mechanisms of urethral fold fusion [13] and epidemiological data indicate that exposure to EDs may be modulators of male urogenital development [14]. Renal anomalies are less common in our cohort (7.7%), but biologically plausible since both the urinary and genital systems have a mesonephric origin of the ducts [7]. Although urogenital anomalies were more frequently observed in children aged 3–7 years compared with those older than 7 years, this difference was not statistically

significant ($p = 0.092$), suggesting that these anomalies are congenital rather than age-dependent, and that the age at presentation may influence their detection.

Ludorf et al. (7) reported patterns of co-occurring birth defects with hypospadias, demonstrating significant clustering of urogenital, musculoskeletal and cardiovascular anomalies, also revealed certain clustering of concomitant defects, whereas Schneuer et al. [17] highlighted the substantial surgical burden associated with hypospadias management. Modern epidemiological and registry-based studies have characterised severity-stratified prevalence patterns and epidemiological trends of hypospadias across different populations [2,18], supporting the necessity of systematic assessment at presentation. Ludorf et al, similarly demonstrated that approximately 21% of infants with hypospadias harbour at least one additional birth defect, with the urogenital system the most frequently affected [7]. In the same vein, Wu et al. similarly identified cryptorchidism as the most frequent co-existing anomaly, present in 9.3% of their hypospadias series [19]. From a clinical standpoint, the high prevalence of associated anomalies particularly cryptorchidism (22.9%) observed in children presenting with penoscrotal and scrotal hypospadias supports the routine adoption of structured pre-operative screening, including bilateral inguinoscrotal examination and renal ultrasonography, in all hypospadias patients referred to tertiary paediatric surgical units in Pakistan.

The strengths of the study are a large sample size and standardised clinical definitions, as well as universal ultrasonographic screening for renal anomalies, which increases uniformity in diagnostics. However, any weaknesses must not be ignored. There is a single-centre design, and the data from this study are not generalisable, and non-probability consecutive sampling may introduce selection bias. Because the cross-sectional nature does not permit consideration of causal correlation or longitudinal effects. Furthermore, hypospadias was not stratified by severity and frequency of the anomaly but analysed statistically to help determine risk gradients.

CONCLUSION

Urogenital anomalies were commonly found in patients who presented with hypospadias, with the most common being undescended testes, followed by inguinal hernias, renal anomalies, and ambiguous genitalia. Younger children (3-7 years old) showed a relatively higher percentage of urogenital anomalies, but there was an insignificant association between age group and the presence of urogenital anomalies. A while highly significant association was noted between the hypospadias type and the presence of urogenital anomalies.

REFERENCES

1. Van der Horst HJ, De Wall LL. Hypospadias, all there is to know. *Eur J Pediatr*. 2017;176(4):435-441.
2. Shan X, Aguilar S, Canon S. Hypospadias prevalence by severity and associated risk factors: a population-based active surveillance analysis. *J Pediatr Urol*. 2023;19(6):720.e1.
3. Halaseh SA, Halaseh S, Ashour M, Ashour ME. Hypospadias: a comprehensive review including its embryology, etiology and surgical techniques. *Cureus*. 2022;14(7):e27071.
4. Chang J, Wang S, Zheng Z. Etiology of hypospadias: a comparative review of genetic factors and developmental processes between human and animal models. *Res Rep Urol*. 2020;12:673-686.
5. Ansari MS, Chakravarthy P, Yadav P. Hypospadias embryology, etiology, and classification. In: *Hypospadiology: Principles and Practices*. Singapore: Springer Singapore; 2022. p. 9-16.
6. Sparks TN, Society for Maternal-Fetal Medicine Publications Committee. Society for Maternal-Fetal Medicine Consult Series #58: Hypospadias. *Am J Obstet Gynecol*. 2021;225(5):B18–20. doi:10.1016/j.ajog.2021.06.023.
7. Ludorf KL, Benjamin RH, Sanchez ML, McLean SD, Northrup H, Mitchell LE, et al. Patterns of co-occurring birth defects among infants with hypospadias. *J Pediatr Urol*. 2021;17(1):64.e1-64.e8.
8. Ahmad S, Ali QA. Frequency of common urogenital anomalies in patients presenting with hypospadias. *Khyber J Med Sci*. 2021;14(3):144-147.
9. Hariyono DC. Profile of hypospadias patients at Dr Moewardi General Hospital Surakarta period 2015-2020. *Int Surg J*. 2022;9(3):530-533.
10. Patra SS, Swain NN. The study of spectrum of hypospadias and evaluation of its associated anomalies in eastern parts of India. *J Evol Med Dent Sci*. 2017;6(34):2798-2800.
11. Snodgrass W, Bush N. Hypospadias. In: Wein AJ et al., editors. *Campbell-Walsh-Wein Urology*. 12th ed. Philadelphia: Elsevier; 2021. p. 745–82.
12. Joodi M, Amerizadeh F, Hassanian SM, et al. The genetic factors contributing to hypospadias and their clinical utility in its diagnosis. *J Cell Physiol*. 2023;238(5):1029–37.
13. Zhang K, Chen Y, Liu L, Lu M, Cheng J, Gao F, et al. The triglycerides and glucose index rather than HOMA-IR is more associated with hypogonadism in Chinese men. *Sci Rep*. 2017;7(1):15874.

14. Albadawi EA, Alzaman NS, Elhassan YH, Eltahir HM, Abouzied MM, Albadrani MS. The association between maternal endocrine-disrupting chemical exposure during pregnancy and the incidence of male urogenital defects: a systematic review and meta-analysis. *Metabolites*. 2024;14(9):477.
15. Mongioi LM, Calogero AE, Vicari E, Condorelli RA, Russo GI, Privitera S, et al. The role of environmental endocrine disruptors in male reproductive function. *Andrology*. 2022;10(5):817–32. doi:10.1111/andr.13207.
16. Pan W, Wu M, Chen Y, Xie H, Huang Y, Chen F. Boys born with hypospadias and fetal growth restriction exhibit shorter anogenital distances: a retrospective cross-sectional study. *Front Pediatr*. 2025;13:1602368.
17. Schneuer FJ, Holland AJ, Pereira G, Bower C, Nassar N. Prevalence, repairs and complications of hypospadias: an Australian population-based study. *Arch Dis Child*. 2015;100(11):1038-1043.
18. Bergman JE, Loane M, Vrijheid M, Pierini A, Nijman RJ, Addor MC, et al. Epidemiology of hypospadias in Europe: a registry-based study. *World J Urol*. 2015;33(12):2159-2167.
19. Wu W-H, Chuang J-H, Ting Y-C, Lee S-Y, Hsieh C-S. Developmental anomalies and disabilities associated with hypospadias. *J Urol*. 2002;168:229–32.
20. Yu X, Nassar N, Mastroiacovo P, Canfield M, Groisman B, Bermejo-Sánchez E, et al. Hypospadias prevalence and trends in international birth defect surveillance systems, 1980–2010. *Eur Urol*. 2019;76(4):482-490.
21. Tariq M. Frequency of associated anomalies in patients with hypospadias at a tertiary care military hospital. *Pak Armed Forces Med J*. 2023;73(Suppl-1):S38–42.
22. Hussain N. Spectrum of urogenital anomalies in children with hypospadias: a tertiary-centre experience. *J Coll Physicians Surg Pak*. 2022;32(8):1023–7.