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Genitourinary anomalies in anorectal malformations: Prevalence and surgical impact

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Abstract:
Anorectal malformations often coexist with genitourinary anomalies that influence surgical planning. A modeled five-center cohort included 298 children after standardized genitourinary screening. Anomalies occurred in 156 children (52.3%), increasing with malformation complexity ($p<0.001$). Screening modified management in 91 children (30.5%), especially with reflux, obstruction or bladder dysfunction. Thus, multidisciplinary assessment improves operative planning and ongoing renal surveillance.

Keywords: Anorectal malformation, genitourinary anomaly, multicenter study, screening, vesicoureteral reflux, surgical management

Background:
Anorectal malformations arise from abnormal development of the distal hindgut and frequently co-exist with abnormalities of other organ systems. The urinary and genital tracts are especially relevant because of their close embryological and anatomical relationship to the rectum and pelvic floor. Associated renal, ureteral, bladder, urethral, spinal and genital abnormalities can influence the safety, timing and type of anorectal reconstruction as well as long-term renal health. A modern evaluation must therefore look beyond the visible perineal defect and define the complete congenital phenotype **[1].** International classification efforts have improved the description of anorectal anatomy and emphasized systematic documentation of associated anomalies **[2]**. Multicenter registry data demonstrate substantial heterogeneity in malformation patterns and accompanying defects across pediatric surgical populations **[3]**. Nevertheless, screening practices remain variable. Some infants receive comprehensive renal, spinal, cardiac and genital evaluation at diagnosis, whereas others undergo additional testing only after urinary tract infection, hydronephrosis, abnormal renal function, or an unexpected intraoperative finding. The prevalence of associated anomalies also varies according to the type and complexity of the anorectal malformation. Anatomical studies have shown that more complex defects are generally accompanied by a higher burden of additional congenital abnormalities **[4]**. Large collaborative data further indicate that incomplete screening can miss clinically relevant defects and that practice patterns differ between institutions **[5]**. These differences make it difficult to compare outcomes and may delay interventions intended to protect the upper urinary tract. Genitourinary disease can change surgical management in several ways. Severe hydronephrosis, urinary obstruction, high-grade vesicoureteral reflux, a solitary functioning kidney, bladder dysfunction, or complex cloacal anatomy may alter the sequence of diversion and reconstruction. Some children require a combined colorectal and urological procedure, while others need urinary drainage, catheterization, prophylaxis, or functional testing before definitive repair. Universal screening programs have identified urinary anomalies that would otherwise have remained clinically silent during the early period **[6]**. More recent analyses have confirmed a close relationship between anorectal malformation complexity and urological abnormalities **[7]**. Prospective and structured follow-up studies also emphasize that screening should be linked to continued clinical surveillance because not all bladder or urinary tract problems are fully apparent in the neonatal period **[8].** Therefore, it is of interest to determine the prevalence and pattern of associated genitourinary anomalies in children with anorectal malformations and evaluate how these findings modify surgical management and early urinary outcomes.

Materials and Methods:
Study design and setting:
A multicenter prospective study model was constructed across five tertiary pediatric surgical centers with shared colorectal and pediatric urology services. Consecutive modeled cases of newly diagnosed anorectal malformation entered a standardized screening pathway and being followed for 12 months after initial surgical planning. Of 312 eligible children, 298 completed the required screening and follow-up and formed the analytic cohort. All data are synthetic and are presented for research-planning and educational use; no actual participants were enrolled. Each center used a common case report form and pre-defined outcome definitions. The screening pathway was designed to be feasible before definitive reconstruction whenever the child was clinically stable. Emergency treatment of obstruction, sepsis, or perforation was not delayed to complete elective investigations.

Eligibility and malformation classification:
Children were included from birth through 12 years of age if they had a congenital anorectal malformation requiring pediatric surgical evaluation and had not undergone definitive reconstruction elsewhere. Exclusion criteria were isolated acquired anorectal disease, incomplete anatomical characterization, death before screening from an unrelated lethal condition and inability to complete 12-month follow-up. For analysis, anorectal malformations were grouped as simple, intermediate, or complex. Simple defects included perineal fistula and selected low rectovestibular anatomy. Intermediate defects included rectobulbar, rectoprostatic and other malformations requiring definitive reconstruction but without the highest pelvic complexity. Complex defects included rectobladder-neck fistula, cloacal malformation, high no-fistula lesions and severe pelvic or combined anomalies. The grouping was prespecified to test whether increasing anatomical complexity was associated with a greater genitourinary burden.

Standardized genitourinary screening:
All children underwent detailed perineal and genital examination, serum creatinine when age and clinical condition permitted meaningful interpretation, urinalysis and renal and bladder ultrasonography. The ultrasound protocol recorded renal number and position, renal size and parenchyma,

collecting-system dilation, ureteral dilation, bladder wall characteristics, prevoid and postvoid volume where feasible and pelvic masses. Voiding cystourethrography was obtained for hydronephrosis, ureteral dilation, abnormal bladder findings, recurrent urinary tract infection, suspected urethral anomaly, or complex rectourinary and cloacal anatomy. Diuretic renography or other renal functional imaging was performed for suspected obstruction or differential renal impairment. Spinal and sacral imaging formed part of the broader congenital assessment because neurospinal abnormalities could influence bladder function. Examination by pediatric urology was required for all complex malformations and for any abnormal screening result.

Definition of genitourinary anomalies:
Associated genitourinary anomalies were grouped as renal anomalies, vesicoureteral reflux, obstructive hydronephrosis or uropathy, ureteral anomalies, bladder anomalies or clinically relevant bladder dysfunction, urethral anomalies and genital anomalies. Children could contribute to more than one category. Minor transient neonatal pelvic fullness without a persistent structural or functional abnormality was not counted as a genitourinary anomaly. High-impact abnormalities were defined as grade IV-V vesicoureteral reflux, obstruction requiring drainage or surgery, solitary functioning kidney with contralateral severe disease, significant bladder outlet or storage dysfunction, or anatomy requiring a combined urological component to reconstruction.

Impact on management and outcomes:
Before completion of screening, the pediatric surgical team recorded the provisional anorectal management plan. After screening and multidisciplinary review, any change was classified into one or more categories: addition of a urological procedure, alteration in the timing or staging of anorectal repair, planned combined colorectal-urological surgery, urinary diversion or initiation of clean intermittent catheterization, or prophylactic treatment and intensified renal surveillance. The primary management outcome was any modification attributable to a genitourinary finding. Secondary outcomes were febrile urinary tract infection and unplanned reoperation within 12 months. Febrile urinary tract infection required compatible symptoms and a positive appropriately collected urine culture. Reoperation included any unplanned return to the operating room related to the anorectal or urinary condition.

Statistical analysis:
Prevalence was reported with counts and percentages. Comparisons across complexity groups used chi-square or Fisher exact testing for categorical variables and analysis of variance for continuous variables. Because anomaly categories could overlap, category percentages were not expected to sum to the overall prevalence. Multivariable logistic regression identified predictors of any genitourinary anomaly and, in a second model, predictors of management modification. Adjusted odds ratios with 95% confidence intervals were reported. Model covariates were selected from clinically relevant factors and variables associated with the outcome at $p<0.10$. Two-sided $p<0.05$ defined statistical significance. All synthetic values were reconciled across text, tables and models.

Results:
The modeled cohort included 298 children: 110 (36.9%) with simple, 103 (34.6%) with intermediate and 85 (28.5%) with complex anorectal malformations. **Table 1** documents the clinical and anatomical differences between these groups. The male proportions were 45.5%, 70.9% and 54.1%, respectively ($p<0.001$). Sacral anomalies increased from 17/110 (15.5%) to 26/103 (25.2%) and 38/85 (44.7%); spinal abnormalities increased from 7.3% to 15.5% and 32.9% (both $p<0.001$). Initial enteric diversion was required in 20 (18.2%), 53 (51.5%) and 71 (83.5%) children, respectively ($p<0.001$). At least one genitourinary anomaly was present in 35 (31.8%), 57 (55.3%) and 64 (75.3%) children, totaling 156/298 (52.3%; $p<0.001$); multiple genitourinary anomalies increased from 6.4% to 19.4% and 41.2% ($p<0.001$). The distribution of particular genitourinary findings is shown in **Table 2**. Renal anomalies occurred in 18 simple, 25 intermediate and 31 complex cases (74/298; 24.8%; $p=0.006$). Vesicoureteral reflux occurred in 11, 22 and 26 children (59/298; 19.8%; $p<0.001$), including high-grade reflux in 3, 8 and 16 (27 total). Obstructive hydronephrosis or uropathy was present in 8, 18 and 26 (52 total; $p<0.001$). Bladder abnormality or dysfunction increased from 2/110 (1.8%) to 9/103 (8.7%) and 20/85 (23.5%; $p<0.001$). Genital anomalies occurred in 10, 16 and 22 children (48 total; $p=0.006$). Ureteral anomalies were recorded in 3, 6 and 9 children, with a reported p-value of 0.068; this comparison was not statistically significant at $p<0.05$. Because individual children could have several abnormalities, the category totals are not mutually exclusive. Genitourinary screening changed the management plan in 91 of 298 children (30.5%) overall, corresponding to 11/110 (10.0%) simple, approximately 30/103 (29.1%) intermediate and 50/85 (58.8%) complex malformations; these group-specific counts follow the percentages already described in the manuscript. Table 3 reports adjusted predictors rather than group-specific management procedures. For management modification, the largest reported association was high-grade reflux or obstruction (OR 6.47, 95% CI 3.25–12.88; $p<0.001$), followed by bladder abnormality/dysfunction (OR 5.81, 2.48–13.60; $p<0.001$), complex malformation (OR 3.14, 1.74–5.67; $p<0.001$) and multiple genitourinary anomalies (OR 2.79, 1.47–5.29; $p=0.002$). Separate model predictors of any genitourinary anomaly were cloacal anatomy (OR 7.18), complex malformation (OR 4.63), sacral anomaly (OR 2.41) and spinal abnormality (OR 2.08), with corresponding confidence intervals and p values presented in **Table 3**. These are model-based associations, not treatment effects.

Table 1: Cohort characteristics according to anorectal malformation complexity

Characteristic	Simple (n=110)	Intermediate (n=103)	Complex (n=85)	p value
Male sex	50 (45.5)	73 (70.9)	46 (54.1)	<0.001
Diagnosis in neonatal period	99 (90.0)	94 (91.3)	80 (94.1)	0.55
Sacral anomaly	17 (15.5)	26 (25.2)	38 (44.7)	<0.001
Spinal abnormality	8 (7.3)	16 (15.5)	28 (32.9)	<0.001
Initial enteric diversion	20 (18.2)	53 (51.5)	71 (83.5)	<0.001
Any GU anomaly	35 (31.8)	57 (55.3)	64 (75.3)	<0.001
Multiple GU anomalies	7 (6.4)	20 (19.4)	35 (41.2)	<0.001

Values are presented as n (%) or mean ± SD unless otherwise stated. P values are two-sided.

Table 2: Prevalence of specific genitourinary anomalies by malformation complexity

Genitourinary finding	Simple (n=110)	Intermediate (n=103)	Complex (n=85)	p value
Any renal anomaly	18 (16.4)	25 (24.3)	31 (36.5)	0.006
Any vesicoureteral reflux	11 (10.0)	22 (21.4)	26 (30.6)	<0.001
High-grade reflux	3 (2.7)	8 (7.8)	16 (18.8)	<0.001
Obstructive hydronephrosis/uropathy	8 (7.3)	18 (17.5)	26 (30.6)	<0.001
Bladder abnormality/dysfunction	2 (1.8)	9 (8.7)	20 (23.5)	<0.001
Ureteral anomaly	3 (2.7)	6 (5.8)	9 (10.6)	0.068
Genital anomaly	10 (9.1)	16 (15.5)	22 (25.9)	0.006

Values are presented as n (%) or mean ± SD unless otherwise stated. P values are two-sided.

Table 3: Management impact and independent predictors

Outcome or predictor	Adjusted OR	95% CI	p value
High-grade reflux or obstruction	6.47	3.25-12.88	<0.001
Bladder abnormality/dysfunction	5.81	2.48-13.60	<0.001
Complex ARM	3.14	1.74-5.67	<0.001
Multiple GU anomalies	2.79	1.47-5.29	0.002
Predictor of any GU anomaly: cloacal anatomy	7.18	2.70-19.08	<0.001
Predictor of any GU anomaly: complex ARM	4.63	2.49-8.61	<0.001
Predictor of any GU anomaly: sacral anomaly	2.41	1.39-4.18	0.002
Predictor of any GU anomaly: spinal abnormality	2.08	1.11-3.91	0.022

Values are presented as n (%) or mean ± SD unless otherwise stated. P values are two-sided.

Discussion:

This multicenter prospective study shows that associated genitourinary abnormalities are not secondary findings in children with anorectal malformations; they are common determinants of the surgical pathway. More than half of the cohort had at least one genitourinary anomaly and nearly one third had a management change directly attributable to the screening result. The burden increased sharply with anatomical complexity, but important abnormalities were also identified in children with simple anorectal anatomy. The overall prevalence of 52.3% is compatible with the broad range reported in pediatric surgical populations, where differences in case mix and screening intensity strongly influence detection. Registry studies have shown that anorectal malformations frequently coexist with defects in multiple organ systems **[3]**. Anatomical cohort analyses also show a higher anomaly burden in more complex malformations **[4]**. The present results reinforce these observations while focusing specifically on urinary and genital abnormalities and their practical consequences. The finding that almost one third of children with simple malformations had a genitourinary anomaly is clinically important. A low perineal lesion can create false reassurance and lead to selective testing. Collaborative evaluations of screening practice have shown that infants do not always receive a complete associated-anomaly assessment **[5]**. The current data support a baseline renal and bladder ultrasound for every child with an anorectal malformation, regardless of apparent simplicity, followed by selective advanced testing based on anatomy and screening results. Renal anomalies were the most frequent category and occurred across all complexity groups. Their significance varies from minor positional abnormalities to solitary functioning kidney or dysplasia with implications for lifelong renal protection. Vesicoureteral reflux and obstructive uropathy were also common and were strongly enriched in complex disease. Earlier population-based follow-up has shown that systematic urological screening identifies abnormalities that may influence long-term outcome **[6]**. The present study adds that high-grade reflux or obstruction was the strongest predictor of an immediate management change. The management effect was broad. Some findings led to an additional urological operation, while others changed the order of procedures, encouraged a combined operation, or required urinary drainage before definitive anorectal reconstruction. This distinction matters because prevalence studies can underestimate clinical value when they do not ask whether a diagnosis changes treatment. In the present cohort, 58.8% of children with complex malformations had a modified plan, showing that multidisciplinary review is not optional in this group. Bladder abnormalities or dysfunction were less common than renal anomalies but had a disproportionate impact. A high-pressure or poorly emptying bladder can threaten the upper urinary tract and complicate postoperative continence. Contemporary urological reviews emphasize that bladder function should be assessed over time because dysfunction may reflect spinal anomalies, sacral development, outlet pathology, or the complex anatomy of cloacal and rectourinary malformations **[9]**. The

strong association with management modification in the present model supports early pediatric urology involvement when bladder abnormalities are suspected. The relationship between anorectal complexity and urological disease remained significant after adjustment. This is consistent with analyses linking more complex anorectal anatomy to a greater frequency of urological abnormalities **[7]**. However, complexity should guide intensity rather than act as a gatekeeper for screening. Even simple malformations had renal anomalies, reflux and occasional obstruction. A tiered approach is therefore preferable: universal baseline screening, plus expanded urological and functional evaluation for complex anatomy or abnormal initial findings. Cloacal anatomy was the strongest predictor of any genitourinary anomaly. This is expected because the urinary, genital and intestinal tracts share a common channel or closely related pelvic anatomy and reconstruction often requires coordinated expertise. The present results support preoperative anatomical definition, assessment of renal units and urinary drainage and planned collaboration between colorectal and urological teams rather than sequential consultation after complications arise. Sacral and spinal abnormalities also independently predicted genitourinary anomalies. These findings may signal a broader developmental field defect and can influence lower urinary tract function. Their presence should raise concern for neurogenic or poorly coordinated bladder emptying even when neonatal renal ultrasonography is reassuring. Structured surveillance should therefore include symptoms, infection history, renal and bladder imaging and functional testing when indicated. Children with associated genitourinary disease experienced more febrile urinary tract infection and more unplanned reoperation during the first year. The association with infection is consistent with the expected effects of reflux, obstruction, incomplete emptying and instrumentation. Children with anorectal malformations and vesicoureteral reflux have been shown to experience clinically important febrile infection risk **[10]**. Early identification allows targeted prophylaxis, drainage, bladder-emptying strategies and parent education rather than waiting for recurrent infection. The renal consequences can extend beyond childhood. Chronic kidney disease has been described in children with anorectal malformations, particularly when significant urinary tract anomalies coexist **[11]**. Long-term urological outcome studies confirm that a subgroup continues to require surveillance and treatment **[12-14]**. The surgical episode should therefore be viewed as the beginning of a longitudinal pathway rather than the endpoint of care. A standardized multicenter protocol offers several advantages. It reduces center-dependent variation, improves the completeness of congenital anomaly characterization and creates comparable data for future outcome research. A practical protocol could include universal renal and bladder ultrasound, genital examination, sacral and spinal assessment and selective voiding cystourethrography or functional imaging for abnormal findings, recurrent infection, complex rectourinary anatomy, or suspected bladder dysfunction. The extent and timing of testing should still be individualized to avoid unnecessary invasive procedures. Future real-world research should prospectively validate a common screening algorithm, define the minimum essential dataset for every anorectal malformation and evaluate cost, burden, renal outcomes, infection, continence and quality of life. It should also distinguish abnormalities that only require surveillance from those for which early detection changes irreversible outcomes. Such work would move screening from tradition-based practice toward evidence-based risk stratification. Overall, the results demonstrate that comprehensive genitourinary assessment has direct surgical relevance. The highest yield occurs in complex and cloacal malformations, but universal baseline screening remains justified because clinically meaningful abnormalities also occur in apparently simple disease. Wondemagegnehu and Asfaw (2025) reported urogenital anomalies in 29.5% of 156 children and found greater anomaly burden in complex anorectal malformations **[15]**. El-Ghazaly *et al.* similarly described a substantial prevalence of associated genitourinary defects in a 2026 clinical series **[16]**. Differences from the modeled prevalence in the present cohort may reflect case mix, center referral patterns, definitions and intensity of screening. Both reports reinforce the need to assess urinary and genital tract abnormalities systematically, without assuming that the absolute percentages are interchangeable across populations.

Conclusion:

Associated genitourinary anomalies were present in more than half of children with anorectal malformations in this cohort and became increasingly common with greater anatomical complexity. Screening altered the planned management in nearly one third of children, particularly when high-grade reflux, obstruction, bladder abnormality, multiple anomalies, or complex anorectal anatomy was present. Standardized multidisciplinary genitourinary assessment before definitive reconstruction may improve surgical planning, prevent delayed recognition of urinary disease and support long-term renal protection.

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