



TREATMENT OF ANORECTAL MALFORMATIONS IN BOYS AT THE MODERN STAGE: A LITERATURE REVIEW

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ABSTRACT

Anorectal malformations (ARM) in boys represent a heterogeneous spectrum of congenital anomalies affecting 3.0-3.5 per 10,000 live births, ranging from simple perineal fistulas to complex rectovesical communications. Accurate diagnosis relies on comprehensive clinical examination, cross-table lateral radiography, ultrasonography, and magnetic resonance imaging, with systematic classification according to the Krickenbeck system guiding therapeutic decisions. Associated anomalies occur in 48-78% of cases, predominantly affecting genitourinary (40%) and spinal (25-34%) systems, necessitating multidisciplinary evaluation. Surgical management strategies have evolved significantly, with posterior sagittal anorectoplasty (PSARP) remaining the gold standard while laparoscopic-assisted anorectoplasty (LAARP) offers a minimally invasive alternative with comparable functional outcomes. The choice between single-stage and staged repair depends on malformation complexity, patient anatomy, and institutional expertise. Postoperative complications include anal stricture (6-50%), rectal prolapse (14-30%), and urological complications, particularly in high-type malformations. Functional outcomes demonstrate voluntary bowel movements in 53-62% of patients, with fecal incontinence affecting 42-48% and constipation in 67%. Long-term quality of life is significantly impacted by continence status, emphasizing the importance of comprehensive bowel management programs and lifelong multidisciplinary follow-up for optimal outcomes

Introduction. Anorectal malformations (ARMs) represent a heterogeneous spectrum of congenital anomalies affecting the distal anus, rectum, and frequently the urogenital tract. The global prevalence of ARMs is approximately 3.0-3.5 per 10,000 live births, with significant geographical variations and male predominance [1]. These complex malformations pose substantial challenges to pediatric surgeons worldwide, requiring specialized expertise and individualized treatment approaches.

In boys, ARMs manifest with diverse anatomical configurations, ranging from relatively simple perineal fistulas to complex high-type defects involving rectourethral or rectoprostatic fistulas. The anatomical classification is critical, as it directly influences both surgical strategy and long-term functional prognosis [2]. Despite advances in surgical techniques and perioperative management, ARMs remain associated with significant morbidity, and their treatment continues to evolve with improved understanding of pelvic anatomy and development of minimally invasive approaches.

The management of boys with ARMs has undergone substantial refinement over recent decades. Contemporary practice emphasizes accurate preoperative diagnosis through advanced imaging modalities, careful selection of surgical approach, and comprehensive multidisciplinary care [3]. The posterior sagittal anorectoplasty (PSARP) remains the gold standard for most defects, although laparoscopic-assisted techniques have emerged as viable alternatives for selected cases. Long-term functional outcomes, particularly bowel continence and quality of life, depend heavily on the initial anatomical defect severity, associated anomalies, and quality of surgical reconstruction [4].

This review examines current approaches to diagnosis, classification, surgical management, and functional outcomes in boys with ARMs, synthesizing recent evidence to provide a comprehensive overview of contemporary practice.

Diagnosis and Classification. Early and accurate diagnosis of anorectal malformations in boys is fundamental to appropriate surgical planning and prognostic assessment. The diagnostic approach encompasses careful clinical examination, advanced imaging modalities, and systematic evaluation for associated anomalies. Most cases are diagnosed within the first 24 hours of life through routine neonatal physical examination, although delayed presentations occasionally occur, particularly in less severe defects [5].

Clinical evaluation begins with meticulous perineal inspection to identify the presence and location of any fistulous opening. In male neonates, observation for meconium passage within the first 24 hours is critical. If meconium is not visualized on the perineum or in the urine after this period, further imaging is warranted. The cross-table lateral radiograph, obtained with the infant in prone position after allowing air to reach the distal bowel, remains a useful initial investigation to assess the distance between the rectal gas shadow and the perineal skin [5].

Modern imaging protocols have substantially refined preoperative assessment. Ultrasound has emerged as a valuable, radiation-free modality during the neonatal period, particularly useful for determining the pouch-perineum distance and evaluating the relationship between the distal rectal pouch and puborectalis muscle. Three sonographic approaches – suprapubic, perineal, and infracoccygeal – each provide complementary information regarding fistula location and sphincter complex development [6]. Magnetic resonance imaging (MRI) offers superior anatomical detail, demonstrating particular value in

delineating complex defects, identifying associated spinal dysraphism, and assessing pelvic musculature. Recent comparative studies suggest MRI provides more comprehensive preoperative information than fluoroscopic colostography, especially in boys with high-type malformations requiring colostomy [7]. The diagnostic accuracy of MRI in detecting genitourinary fistulas and associated anomalies makes it increasingly the imaging modality of choice in specialized centers.

Classification systems have evolved to facilitate standardized communication and outcome comparison. The Krickenbeck classification, established in 2005, has become the international standard for categorizing ARMs based on anatomical features rather than the historical "high-low-intermediate" terminology. For male patients, this classification distinguishes perineal fistula, rectourethral fistula (subdivided into bulbar and prostatic types), rectovesical fistula, and anorectal malformation without fistula [8]. This anatomically precise classification correlates more reliably with functional prognosis and guides surgical approach selection. European consensus guidelines have further refined diagnostic protocols, emphasizing the importance of systematic screening for associated anomalies across all ARM types regardless of severity [8].

Associated congenital anomalies occur in approximately 48-78% of boys with ARMs, necessitating comprehensive evaluation [9]. Genitourinary anomalies represent the most frequent association, present in up to 40% of cases and particularly common in rectourethral and rectovesical fistulas. Spinal dysraphism affects approximately 25-34% of patients, with higher prevalence in complex malformations. Recommended screening includes renal ultrasonography, voiding cystourethrography when indicated, echocardiography for cardiac assessment, and spinal ultrasonography or MRI to detect tethered cord and vertebral anomalies [5,9]. This systematic approach to diagnosis and classification enables precise surgical planning and realistic counseling regarding long-term functional expectations.

Surgical Management. The surgical approach to anorectal malformations in boys must be individualized based on the anatomical defect type, associated anomalies, and institutional expertise. Contemporary management strategies range from single-stage neonatal repairs for low-type defects to staged procedures involving colostomy formation for complex malformations. The evolution of surgical techniques over recent decades has introduced both traditional open approaches and minimally invasive alternatives, each with specific indications and outcomes.

For low-type malformations, particularly perineal fistulas, primary repair without protective colostomy has demonstrated excellent outcomes. A large retrospective analysis of **primary posterior sagittal anorectoplasty (P-PSARP)** performed over two decades showed that male neonates with perineal fistulas could undergo safe primary repair within the first postnatal week, achieving satisfactory functional outcomes with minimal complications [10]. However, intermediate and high-type malformations typically require a staged approach beginning with colostomy creation. The traditional divided sigmoid colostomy remains the preferred diversion method in most specialized centers, as it effectively prevents overflow into the distal limb and reduces urinary tract infections. Recent evidence suggests that loop colostomies, when properly constructed, offer comparable safety with technical simplicity and easier closure, reporting complication rates of approximately 12% [11].

Posterior sagittal anorectoplasty, introduced by Peña in 1982, remains the gold standard surgical technique for repairing ARMs in boys. The PSARP approach provides excellent anatomical exposure of the sphincter complex and allows precise identification and division of rectourethral or rectovesical fistulas under direct vision [12]. The procedure involves a posterior midline sagittal incision, complete division of the muscle complex to expose the rectum, meticulous fistula dissection and ligation, and reconstruction of the anal canal within the sphincter mechanism. Optimal timing for definitive repair is typically between 1-6 months of age, allowing adequate infant growth while avoiding prolonged colostomy-related morbidity [13]. Postoperative anal dilations, once considered mandatory, have been challenged by recent randomized controlled trials demonstrating that routine dilations do not significantly reduce stricture formation, with rates remaining around 10-13% regardless of dilation protocols [14]. However, standardized dilation protocols when employed appear to minimize stricture complications, with reported rates as low as 7.7% in large series [15].

Laparoscopic-assisted anorectoplasty (LAARP) has emerged as an important minimally invasive alternative to PSARP since its introduction by Georgeson in 2000. The laparoscopic approach offers theoretical advantages including preservation of the levator ani muscle complex, reduced posterior dissection and scarring, superior visualization of high rectal pouches, and smaller perineal wounds [16]. The technique involves laparoscopic mobilization of the rectum, intracorporeal fistula division, creation of a pull-through tract under electrostimulator guidance, and perineal anoplasty [17]. Multiple comparative studies have evaluated LAARP versus PSARP outcomes. A systematic review and meta-analysis of 254 patients demonstrated that LAARP achieved significantly shorter hospital stays compared to PSARP (10.9 vs 14.4 days, $p < 0.0001$), with no significant differences in total postoperative complications [18]. Recent multi-center data from five tertiary institutions involving 136 male patients with rectoprostatic and rectobulbar fistulas found comparable functional outcomes between LAARP and PSARP, with VACTERL association and fistula location being more significant prognostic factors than surgical approach [4].

However, LAARP is not without specific complications. Rectal mucosal prolapse occurs more frequently after LAARP than PSARP, with reported incidences reaching 9-18% in some series, though this can be mitigated by suture fixation of the rectum to presacral fascia [19]. Posterior urethral diverticulum (PUD), a potentially serious complication, has been reported following LAARP, particularly before the introduction of routine intraoperative urethroscopy to guide fistula division [20]. Contemporary techniques incorporating urethroscopic guidance have significantly reduced PUD occurrence. The debate regarding LAARP suitability for rectobulbar versus rectoprostatic fistulas continues, with some centers advocating PSARP for bulbar fistulas due to concerns about adequate distal dissection [21].

For high-type malformations requiring extensive abdominal mobilization, combined laparoscopic and perineal approaches offer distinct advantages. Modified techniques include transumbilical colostomy creation followed by simultaneous colostomy closure and LAARP, providing excellent cosmetic results and avoiding additional abdominal scars [22]. In cases where the distal colostomy segment is severely shortened or abnormal, laparoscopic-assisted distal colon excision with proximal colon pull-through anorectoplasty (PCPARP) has shown comparable functional outcomes to standard LAARP [23].

Single-stage primary repair without protective colostomy has gained acceptance for selected patients. Neonatal P-PSARP for intermediate-type malformations where the rectal pouch reaches the ossified fifth sacral vertebra on invertogram has demonstrated feasibility and safety, avoiding colostomy-related morbidity [24]. A comparative study of single-stage versus staged PSARP suggested that primary repair may be safe and feasible even for high-type ARMs in appropriate candidates, potentially allowing early restoration of normal defecation patterns [18]. However, patient selection remains critical, with exclusion criteria typically including low birth weight (<2.5 kg), significant associated anomalies, and rectovesical fistulas.

Surgical decision-making must account for institutional experience and surgeon expertise. Centers with high-volume ARM experience report superior outcomes regardless of technique, emphasizing the importance of centralized care [25]. The choice between PSARP and LAARP should consider the specific fistula type, surgeon familiarity, and available resources rather than dogmatic adherence to one approach [26]. Both techniques, when performed meticulously with attention to anatomic principles, can achieve satisfactory functional outcomes in appropriately selected patients.

Complications and Functional Outcomes. Postoperative complications following anorectal malformation repair in boys encompass both early surgical issues and long-term functional sequelae that significantly impact quality of life. Understanding the spectrum of complications and their management is essential for optimizing patient outcomes and setting realistic expectations for families.

Anal stricture represents one of the most common early complications, occurring in 6-50% of cases depending on surgical technique and postoperative management protocols. Traditional PSARP demonstrates stricture rates of approximately 6-8%, while laparoscopic approaches report higher incidences ranging from 30-50% when tunnels through the sphincter complex are inadequately sized [27]. Standardized postoperative dilation protocols have been advocated to prevent strictures, though recent evidence questions their necessity, with some studies reporting comparable stricture rates of 7.7% with or without routine dilations [28]. Strictures occurring at the skin level can be effectively managed with Heineke-Mikulicz anoplasty, while longer proximal strictures may require redo pull-through procedures with mobilization of healthy proximal bowel [29].

Rectal prolapse develops in 14-30% of patients following ARM repair, with increased incidence observed in boys with high-type malformations, sacral abnormalities, and poorly developed pelvic musculature. Rectourethral prostatic and rectovesical fistulas demonstrate the highest prolapse rates at 29% and 29% respectively [30]. While many cases of mucosal prolapse can be managed conservatively, approximately 68% require operative intervention. Prolapse repair itself carries significant morbidity, with 31% developing postoperative strictures and 31% experiencing recurrent prolapse requiring repeat procedures [31].

Urological complications occur predominantly in males due to the intimate relationship between the rectum and urethra. Urethral injury during fistula division can lead to stricture formation, posterior urethral diverticulum (remnant of the original fistula), or persistent fistula. These complications may present with recurrent urinary tract infections, voiding dysfunction, or difficulty with catheterization. Neurogenic urinary retention affects a subset of patients, particularly those with associated spinal dysraphism, requiring clean intermittent catheterization and occasionally bladder augmentation [32].

Functional outcomes vary considerably based on malformation type, surgical quality, and presence of prognostic factors. Comprehensive assessment using the Krickenbeck classification evaluates three key parameters: voluntary bowel movement (VBM), soiling, and constipation. Recent multi-center data demonstrate that 53-62% of patients achieve VBM, while fecal incontinence affects 42-48% of the ARM population, with severe incontinence (Krickenbeck grades 2-3) occurring in 31-37% [29,33]. Constipation represents a major functional challenge, reported in 67% of patients, with prolonged colonic transit times particularly in the rectosigmoid region [34]. Patients with refractory constipation may develop megarectosigmoid, contributing to overflow incontinence and potentially requiring sigmoid resection.

Quality of life studies reveal that children with poor fecal continence experience significant psychosocial impact, including problems in peer relationships (17%), school absences (18%), dietary restrictions (24%), and behavioral difficulties (67% versus 9% in continent patients) [28]. Long-term assessments demonstrate that ARM patients have significantly lower quality of life scores compared to healthy controls, with 57% exhibiting poor bowel function and 32% demonstrating voiding dysfunction requiring ancillary aids [28]. Adult male patients report ongoing concerns regarding sexual function, with erectile and ejaculatory dysfunction documented in this population [35].

Anorectal manometry findings correlate with functional outcomes, demonstrating that sphincter defects, reduced squeeze pressures, and abnormal sacral ratios predict worse continence results. Interestingly, assessment of VBM initially may be deceptive, as patients receiving appropriate laxative treatment show improvement in soiling scores more conspicuously than constipation scores [34]. Redo operations for anatomical complications (mislocation, stricture, persistent fistula) can restore continence in selected patients, with 76% of those with good sphincter quality achieving complete continence following reoperation [36]. However, realistic counseling regarding the high incidence of fecal incontinence and the potential need for comprehensive bowel management programs is essential for appropriate family expectations and long-term psychosocial adjustment.

Conclusion

Anorectal malformations in boys represent a complex spectrum of congenital anomalies requiring individualized surgical management and comprehensive long-term care. Accurate diagnosis through physical examination, imaging modalities, and systematic classification using the Krickenbeck system forms the foundation for optimal treatment planning and prognostic assessment. The high incidence of associated anomalies, particularly affecting the genitourinary and spinal systems, necessitates thorough preoperative evaluation and multidisciplinary collaboration.

Contemporary surgical approaches, including both traditional posterior sagittal anorectoplasty and laparoscopic-assisted techniques, have evolved significantly over recent decades. While PSARP remains the gold standard with proven long-term outcomes, LAARP offers a minimally invasive alternative with comparable functional results when performed by experienced surgeons. The choice between single-stage and staged repair must be individualized based on malformation complexity, patient anatomy, and institutional expertise. Both approaches achieve satisfactory outcomes when anatomical principles are respected and meticulous surgical technique is employed.

Despite surgical advances, functional challenges persist, with approximately half of patients experiencing some degree of fecal incontinence and many requiring bowel management programs. Postoperative complications including stricture, prolapse, and urological dysfunction require ongoing surveillance and intervention. Quality of life considerations extend beyond physical function to encompass psychosocial adjustment and sexual health in adolescence and adulthood.

Future progress depends on centralization of care in high-volume centers, refinement of surgical techniques to minimize complications, development of standardized bowel management protocols, and continued research into predictive factors for long-term outcomes. Comprehensive, lifelong multidisciplinary follow-up remains essential to optimize functional results and quality of life for this vulnerable patient population.

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