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Rectal Atresia in Children: A Comprehensive Review of Recent Advances (2022-2025)

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Abstract

Rectal atresia is a rare congenital anomaly characterized by complete obstruction of the distal rectum, requiring timely surgical intervention. This review synthesizes the latest evidence (2022-2025) on epidemiology, pathogenesis, diagnostic strategies, surgical techniques, postoperative management, and long-term outcomes. The discussion highlights advancements in minimally invasive surgery, genetic research, and patient-centered care, supported by 26 recent studies.

Keywords: Rectal Atresia; Anorectal Malformations (ARMs); Minimally Invasive Surgery; Prenatal Diagnosis; Postoperative Complications

Abbreviations: ARMs: Anorectal Malformations; VACTERL: Vertebral, Anal, Cardiovascular, Tracheoesophageal, Renal, and Limb Anomalies; HOX: Homeobox; WNT: Wingless-Integrated; GLI2: GLI Family Zinc Finger 2; BMP4: Bone Morphogenetic Protein 4; L-PSARP: Laparoscopic-assisted Posterior Sagittal Anorectoplasty; RCT: Randomized Controlled Trial; TEM: Transanal Endoscopic Microsurgery; MCA: Magnetic Compression Anastomosis; PedsQL™: Pediatric Quality of Life Inventory

Epidemiology and Classification

Rectal atresia accounts for 1-2% of anorectal malformations (ARMs), with a male predominance [1]. It often presents as an isolated defect or in association with syndromic conditions such as VACTERL (vertebral, anal, cardiovascular, tracheoesophageal, renal, and limb anomalies) [2-4]. Recent studies emphasize the importance of prenatal diagnosis through ultrasound, which can detect bowel dilation or absent anal dimple as early as the second trimester [2]. Postnatal evaluation includes contrast enema and MRI to confirm the atresia level and rule out fistulas [2,5].

Pathogenesis and Genetics

The etiology of rectal atresia remains multifactorial, involving genetic and environmental factors. HOX gene mutations and disruptions in the WNT signaling pathway during hindgut development are key drivers [6,7]. Recent studies have identified variants in GLI2 and BMP4 as potential genetic risk factors, linking them to altered epithelial-mesenchymal interactions [7]. Animal models demonstrate that maternal obesity and hyperglycemia may disrupt fetal gut vasculature, contributing to atresia [6].

Diagnostic Innovations

Prenatal ultrasound remains the primary screening tool, with 3D imaging enhancing visualization of rectal anatomy [2,5]. Postnatally, contrast enema and MRI are critical for determining the atresia gap and fistula presence [2,5]. Rectal manometry and endoanal ultrasound assess sphincter function, guiding surgical planning [8,9].

Surgical Management

Minimally Invasive Approaches

Laparoscopic-assisted posterior sagittal anorectoplasty (L-PSARP) has gained traction, offering reduced blood loss, shorter hospital stays, and improved cosmetic outcomes compared to open surgery [10,11]. A 2023 randomized controlled trial (RCT) reported a 15% complication rate with L-PSARP versus 35% with open PSARP, highlighting its safety [11]. Transanal endoscopic microsurgery (TEM) is increasingly used for low-type atresia, achieving anastomosis with minimal dissection [12,13].

Fistula Management

Complex cases with rectovesical or rectovaginal fistulas require multi-stage repair. Laparoscopic-assisted fistula ligation combined with transanal anastomosis has shown promising results, preserving sphincter function [10,11].

Emerging Techniques

Robotic surgery is being explored for high-type atresia, offering enhanced precision in pelvic dissection [13]. Magnetic compression anastomosis (MCA) has demonstrated feasibility in reducing anastomotic tension, though long-term data are limited [13].

Postoperative Complications and Management

Anastomotic Stenosis

Stenosis occurs in 5-10% of cases, managed with serial dilatations or endoscopic balloon dilation [14,15]. A 2024 study reported a 90% success rate with endoscopic incision for refractory stenosis [15].

Fecal Incontinence

Long-term incontinence affects 20-30% of patients, linked to sphincter injury during surgery [8,16]. Biofeedback therapy and sacral nerve stimulation show efficacy in improving continence scores [16,17].

Enterocolitis

Enterocolitis, a life-threatening complication, is managed with antibiotics, rectal irrigation, and temporary stoma revision [14]. Probiotic supplementation may reduce recurrence rates [14].

Long-Term Outcomes and Quality of Life

Functional Outcomes

Most patients achieve bowel control by school age, though constipation and soiling remain common [16,18]. A 2023 study reported 80% fecal continence at 5 years post-surgery, with laparoscopic approaches yielding better outcomes [11].

Quality of Life (QOL)

QOL assessments using PedsQL™ show reduced scores in patients with incontinence or stomas [16,19-22]. Psychological support and family counseling are integral to addressing social and emotional challenges [16,19].

Fertility and Sexual Function

Male patients may experience ejaculatory dysfunction, while females may face vaginal stenosis or infertility [17,18]. Multidisciplinary follow-up is recommended to address these issues [17,18].

Future Directions

- Genetic Counseling: Identifying high-risk families through genetic testing for HOX and WNT pathway mutations [6,7].
- Stem Cell Therapy: Preclinical studies explore stem cell-based regeneration of rectal tissue [17].
- AI-Driven Diagnostics: Machine learning algorithms for prenatal ultrasound analysis to improve early detection [5,17].

Conclusion

Rectal atresia management has evolved significantly, with minimally invasive surgery and genetic research driving improvements in outcomes. Multidisciplinary care, including psychological support and long-term follow-up, is essential to optimize patient quality of life. Future advancements in molecular diagnostics and regenerative medicine hold promise for further enhancing care.

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