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A Rare Case of Rectal Atresia: Succesful Management and The Relevance of Follow Up

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ABSTRACT

Background: Rectal atresia (RA) is an uncommon congenital anorectal malformation, accounting for approximately 1–2% of such anomalies. It is characterized by a normally developed anus with a complete or partial discontinuity in the rectal lumen, leading to obstruction. Early identification is often delayed due to a normal perineal appearance, with typical presentation including failure to pass meconium, abdominal distension, and signs of intestinal obstruction. Surgical correction, commonly through posterior sagittal anorectoplasty (PSARP), is the mainstay of treatment, and long-term follow-up is crucial for managing complications and associated anomalies.

Case Presentation: We report the case of a male neonate, diagnosed with RA within 12 hours of birth, presenting with failure to pass meconium and abdominal distension. Despite a normally formed anus, rectal catheterization revealed obstruction 2 cm from the anal verge. Imaging identified left crossed fused renal ectopia. A staged surgical repair was performed: high sigmoid colostomy on day 2, PSARP at 5 months, and colostomy closure at 1 year. Postoperative follow-up revealed recurrent febrile UTIs and constipation. Investigations showed bilateral vesicoureteric reflux and a cross-fused ectopic kidney. The patient underwent ureteric reimplantation at 5 years. At 10 years of age, the child remains continent, infection-free, and demonstrates normal growth and development.

Conclusion: This case highlights not only the successful surgical management of RA but also the importance of long-term follow-up for early detection and management of associated congenital anomalies, particularly urological abnormalities. It underscores the necessity for a multidisciplinary approach in rare anorectal malformations to ensure optimal functional outcomes and quality of life.

Keywords: *Rectal Atresia, Anorectal Malformation, Posterior Sagittal Anorectoplasty (PSARP), Crossed Fused Renal Ectopia, Vesicoureteric Reflux (VUR), Congenital Anomalies, Neonatal Intestinal Obstruction*

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1. INTRODUCTION

Rectal atresia (RA) is a rare type of congenital anorectal malformation characterized by the presence of a normally developed anus but a luminal discontinuity in the rectum, leading to an obstruction to the passage meconium aborally. It accounts for approximately 1-2% of anorectal malformations and can range in spectrum from a simple membranous obstruction to a complete atresia with completely disconnected rectal segments[1]. Newborns with RA typically present with symptoms such as failure to pass meconium, abdominal distension, vomiting and other signs of intestinal obstruction.[2]

Despite a normally appearing anus, passage of rigid rubber tube will reveal a blind-ending rectal pouch which can be demonstrated by inability to pass red rubber tube of 6 No. beyond 1.5 cms to 3 cms per rectally. Imaging techniques, including X-rays, contrast enemas, and MRI, play a crucial role in confirmation of the diagnosis and also dictate management protocols. A contrast enema often reveals a blind rectal pouch with an atretic segment.[3] The exact cause of rectal atresia remains uncertain, though in utero ischemic events have been suggested as a most possible explanation. Unlike other anorectal malformations, the anal sphincter muscle complex and perineal structures are typically well-developed.[4-6] Treatment is surgical and involves restoring intestinal continuity while preserving bowel control. Several surgical approaches have been described, including: posterior sagittal anorectoplasty, transanal pull-through procedures, which allow for direct access to the rectal atresia and minimize external scarring. The most accepted Posterior sagittal anorectoplasty (PSARP) approach , provides better visibility of the rectal, sphincter muscle complex anatomy and helps in a more extensive dissection and end-to-end anastomosis, giving best results when the rectal segments are closely aligned.[2] With timely and appropriate surgical intervention, most infants achieve normal continence. However, long-term issues such as chronic constipation or incontinence can occur. The choice of surgical technique plays a crucial role in preserving anorectal function and preventing complications.

Classification of rectal atresia

Sharma and Gupta Classification:

Type I: Rectal Stenosis: The rectum has a narrowed or constricted passage. This can be further divided into intramural (within the rectal wall) or web-like (a membrane partially obstructing the passage).

Type II: Rectal Atresia with Septal Defect: A membrane or septum separates the proximal and distal rectum, but there is a small opening or communication between the two segments.

Type III: Rectal Atresia with Fibrous Cord: The rectum ends in two blind pouches, with a fibrous cord connecting the two ends.

Type IV: Rectal Atresia with Gap: A gap exists between the two blind pouches of the rectum, with no connection or fibrous cord.

Type V: Multiple Rectal Atresia/Stenosis: This includes various combinations, such as rectal atresia with stenosis, multiple rectal atresias, or thickened rectal valves.

Dorairajan Classification

Grade 1: Short gap between the proximal and distal rectum.

Grade 2: Long gap between the proximal and distal rectum.

Grade 3: Membranous septal type II.

Grade 4: Rectal stenosis

These classifications help guide the choice of surgical treatment based on the severity of the atresia. [2,7]

2. CASE REPORT PRESENTATION

A 12-hour-old male neonate, born to a primigravida mother at 37 weeks of gestation via normal vaginal delivery, was brought to the casualty department with complaints of failure to pass meconium since birth and progressive abdominal distension. The antenatal period was uneventful. Patient also had a minute coloboma in left eye iris level.

Clinical Examination

The baby was alert and active, with a normal cry, good muscle tone, and stable vital signs. Abdominal examination revealed a distended, shiny abdomen. Despite the anus appearing externally normal, a red rubber catheter (size 6) could not be advanced beyond 2 cm from the anal verge, raising suspicion of rectal atresia. A notable additional finding was a small coloboma of the iris in the left eye, indicating a possible syndromic or multisystem involvement.

Investigations

Initial imaging with ultrasonography of the abdomen revealed mild dilation of the pelvicalyceal system on the right kidney. Interestingly, the left kidney was not visualized in its usual anatomical location. Instead, it was found fused to the lower pole of the right kidney and situated on the right side, indicating a diagnosis of left crossed fused renal ectopia. Further investigations were initiated at two years of age when the child developed recurrent febrile urinary tract infections. A micturating cystourethrogram (MCUG) demonstrated grade IV vesicoureteric reflux (VUR) on the right side and grade II reflux on the left, in association with the previously identified renal anomaly. Cystoscopy performed subsequently revealed a mildly trabeculated bladder and widely spaced ureteric orifices, consistent with longstanding high-pressure urinary reflux. To assess renal cortical function and scarring, a DMSA renal scan was performed, which confirmed reduced cortical

function in the ectopic left kidney and fusion with the right kidney's lower pole.

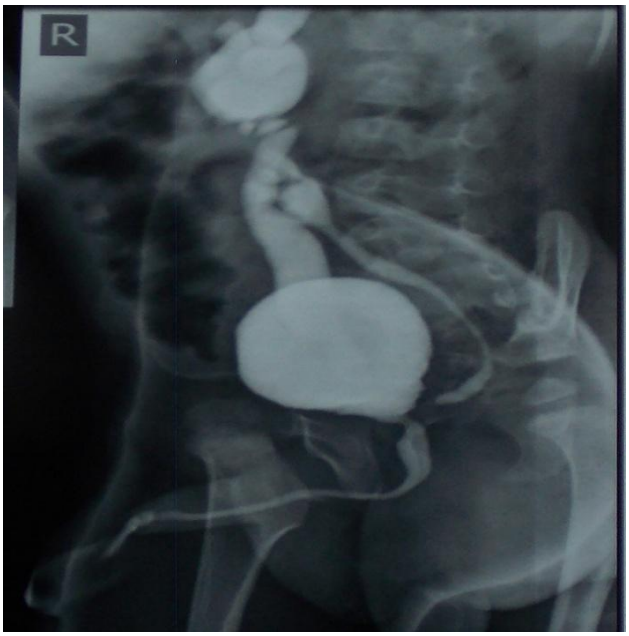

Fig 1: MCU showing right sided Grade IV VUR and left side showing Grade II VUR

Diagnosis

Based on clinical, radiological, and intraoperative findings, the neonate was diagnosed with Type III rectal atresia, characterized by two blind rectal ends connected by a fibrous cord. Associated anomalies included left crossed fused renal ectopia, bilateral vesicoureteric reflux, and left iris coloboma. These multisystem anomalies highlighted the need for a multidisciplinary approach and long-term follow-up.

Management

In the neonatal period, a staged surgical approach was adopted. On the second day of life, the child underwent a high sigmoid colostomy to decompress the bowel and prevent further complications from distal intestinal obstruction. At five months of age, definitive surgical correction was undertaken using the posterior sagittal anorectoplasty (PSARP) technique, which allowed precise dissection and end-to-end anastomosis of the rectal pouches while preserving the sphincter mechanism. At one year of age, following successful neo-anal calibration and assessment of continence potential, the colostomy was closed. The child recovered uneventfully from all surgical stages. At two years of age, due to persistent constipation and recurrent urinary tract infections, further urological evaluation revealed significant bilateral VUR with associated renal anomalies. The child was placed on antibiotic prophylaxis; however, breakthrough infections necessitated surgical intervention. At five years of age, bilateral ureteric reimplantation was performed.

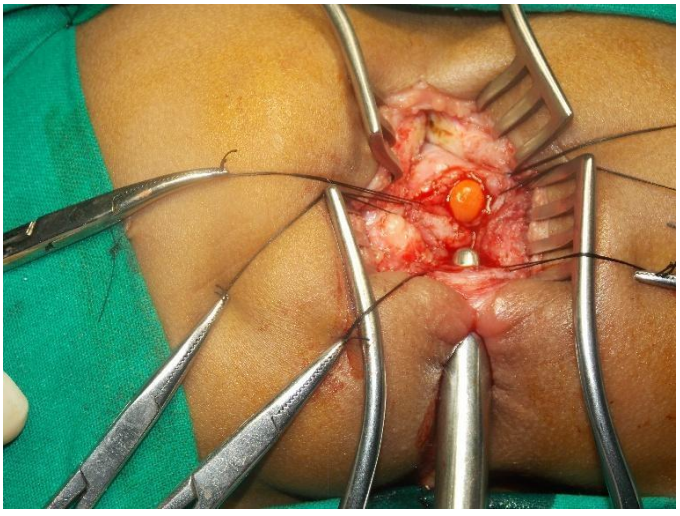

Fig 2: Posterior sagittal Anorectoplasty

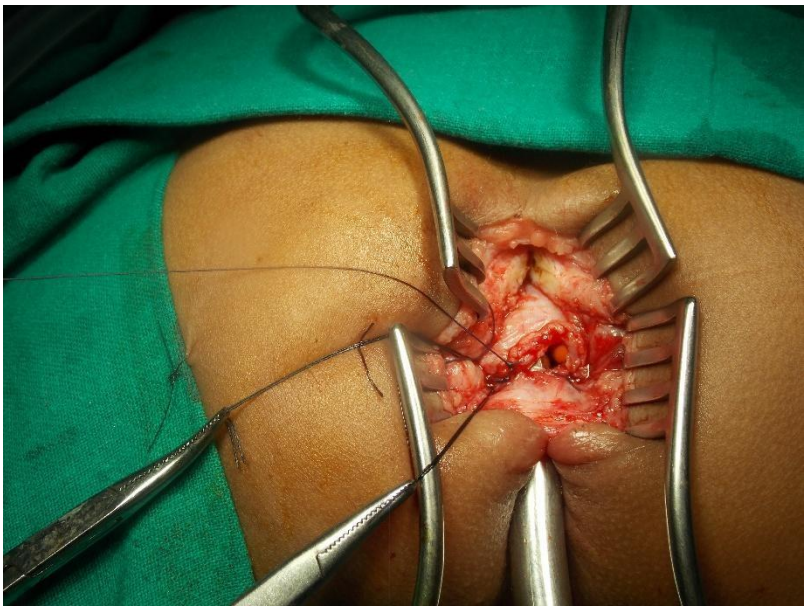

Fig 3: Post completion of surgery

Follow-Up and Outcome

The child has been under regular follow-up and is currently ten years old. Postoperative outcomes have been favourable, with the child demonstrating good continence scores and minimal or no soiling. There have been no further episodes of urinary tract infections following the ureteric reimplantation. The renal function has remained stable, and the child's growth and development are within normal limits for age. The iris coloboma has not resulted in any significant visual impairment. This case highlights the importance of long-term monitoring in patients with rectal atresia, particularly in the presence of associated genitourinary anomalies, to ensure timely intervention and favourable long-term outcomes.

3. DISCUSSION

The case report describes a male newborn diagnosed at birth to have anorectal malformation, presenting with abdominal distension and failure to pass meconium.[1] Our case is similar to other reported cases in literature, which resembles in most aspects with a combination of multi-system anomalies which were extensive, and were diagnosed and treated appropriately and timely as well. [1,2] As in most cases, where RA is usually diagnosed in the neonatal period due to delayed meconium passage and intestinal obstruction despite a normally developed and visible normal looking anusClick or tap here to enter text.[3,4]. The case also mentions an associated congenital renal anomalies, namely crossed renal ectopia, presence of bilateral vesicoureteric reflux, coloboma of iris, whereas literature indicates that RA is often associated with vertebral, cardiac, renal, and spinal anomalies, though specific renal anomalies like crossed ectopia are rarely highlighted.[5]

The case describes a staged surgical approach, with a high sigmoid colostomy performed at birth, followed by posterior sagittal anorectoplasty (PSARP) surgery and colostomy closure at 1 year and 5 months respectively. [5,6] This aligns with literature stating that PSARP is one of the most commonly performed procedures for RA correction. However, some studies suggest alternative surgical techniques, such as transanal end-to-end rectoanal anastomosis(TERA), a pull through surgeries ike Duhamel's or Soave's procedure, and laparoscopy assisted surgeries. which may minimize scarring and provide better visualization.[3,4] Unlike this case report, which documents a long-term follow-up of 12 years, most studies focus on short- to mid-term outcomes, often within 1 to 5 years. [5,7] Another key difference is the postoperative complications noted in this case, specifically chronic constipation and recurrent urinary tract infections due to vesicoureteric reflux.[5,7] While chronic constipation is a well-documented issue in RA cases, urinary complications are less frequently reported in the literature unless there is an associated urogenital anomaly. [5] The case also has multiple post-surgical interventions, including use of dilators for neo anal dilatation, micturiting cystourethrograms, cystoscopy and reimplantation for bilateral vesicoureteric reflux, and observation for coloboma as was not causing major issues with vision, highlighting a more complex and multi system long-term management plan than typically discussed in studies that primarily focus on stool continence. [2,5,8]

In summary, while the case report shares similarities with the existing literature in terms of RA classification and surgical management, it presents a unique perspective due to the presence of a congenital renal anomaly, a longer follow-up period, the need of evaluation and for additional urological interventions. The case also emphasizes the importance of long-term monitoring and interdisciplinary management, which is not always extensively discussed in standard RA studies[3, 7] leading

to a near normal growth and development in these children as they grow into adults with good transition.

4. CONCLUSION

Rectal atresia is a rare anorectal malformation, marked by a normally formed anal canal without continuity to the proximal rectum. Diagnosis may be delayed due to a normal perineal appearance and is confirmed through clinical examination and contrast imaging. The severity of the anomaly varies, guiding surgical management. Among available techniques, posterior sagittal anorectoplasty is widely favored for preserving continence and achieving good outcomes. Given the potential for associated anomalies and long-term complications, early diagnosis, tailored surgical planning, and extended follow-up are crucial for optimal prognosis.

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