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Anal atresia: A report on 10 cases

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Abstract

Introduction: Anal atresia is a congenital anorectal malformation characterized by the complete absence of an opening connecting the rectum to the skin of the perineum. The diagnosis should be made at birth, during the newborn's examination in the delivery room. If this does not occur, the newborn will present with symptoms of intestinal obstruction, including abdominal distension and bilious vomiting.

Materials and Methods: Our retrospective study includes 10 cases of anal atresia treated in 2025 at the pediatric neonatology unit of the Rabat Children's Hospital.

Results: The newborns were predominantly male; 9 out of 10 pregnancies were well managed and carried to term, with one preterm birth at 36–37 weeks' gestation; 9 out of 10 pregnancies resulted in vaginal deliveries, with birth weights ranging from 2000 g to 5000 g; and Apgar scores were normal in half of the cases. The medical history revealed no history of urogenital infection during the first trimester in the mothers, nor any history of exposure to teratogenic medications or tobacco or alcohol use during that period. Furthermore, there is no history of congenital malformations in the family. However, a case of third-degree consanguinity was reported in a single mother with a history of gestational diabetes who was on a diet.

Our patients were admitted between 10 hours and 5 days of life; one-third were admitted due to low neonatal obstruction associated with high anorectal malformation. There was one case of meconium passage through the vagina and four postoperative cases (including two cases with respiratory distress), as well as one preoperative case requiring correction of hyponatremia.

Three out of 10 cases presented with abdominal distension, one case with an ectopic testicle, one case of ambiguous genitalia, and one case with an absent urethral meatus; one newborn presented with trisomic facial features. The malformation screening identified cardiovascular anomalies in half of the patients. Transfontanellar ultrasound was normal in 9 out of 10 cases, and renal ultrasound showed no abnormalities. They received broad-spectrum antibiotic therapy, intravenous fluid replacement, and parenteral analgesia.

Conclusion: This condition is routinely diagnosed in the delivery room. Its severity lies in the presence of associated anomalies. Medical management is part of a multidisciplinary approach that includes medical, surgical, radiological, and genetic care.

Keywords: Anal Atresia; Anorectal Malformation; Diagnosis At Birth; Intestinal Obstruction

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1. Introduction

Anal atresia, or anorectal malformations (ARM), are rare congenital anomalies affecting approximately 1 in 3000 to 5000 births. They are characterized by the absence or abnormal development of the anal opening, requiring neonatal surgical intervention. They are associated with other malformations in approximately 70% of cases. They are very rarely diagnosed prenatally. Imaging plays a crucial role in neonatal care to clarify the anatomy of MAR in cases where clinical diagnosis is difficult and to identify associated malformations.

2. Materials and Methods

Our retrospective study includes 10 cases of anal atresia treated in 2025 at the pediatric neonatology unit of the Rabat Children's Hospital.

3. Results

The newborns were predominantly male; 9 out of 10 pregnancies were well managed and carried to term, with one preterm birth at 36–37 weeks' gestation; 9 out of 10 pregnancies resulted in vaginal deliveries, with birth weights ranging from 2000 g to 5000 g; and Apgar scores were normal in half of the cases. The medical history revealed no history of urogenital infection during the first trimester in the mothers, nor any history of exposure to teratogenic medications or tobacco or alcohol use during that period. Furthermore, there is no history of congenital malformations in the family. However, a case of third-degree consanguinity was reported in a single mother with a history of gestational diabetes who was on a diet.

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On physical examination, the patient's general condition was unremarkable and vital signs were within normal limits. Three out of 10 cases presented with abdominal distension, one case with an ectopic testicle, one case of ambiguous genitalia, and one case with an absent urethral meatus; one newborn presented with trisomic facial features. The malformation screening identified cardiovascular anomalies in half of the patients. Transfontanellar ultrasound was normal in 9 out of 10 cases, and renal ultrasound showed no abnormalities

During the postoperative period, patients were cared for by a multidisciplinary team comprising intensivists and pediatric surgeons. They received broad-spectrum antibiotic therapy, intravenous fluid replacement, and parenteral analgesia.

4. Discussion

Historically referred to as "anal atresia," the international scientific community now prefers the term anorectal malformation (ARM). This term reflects the reality of a broad spectrum of anatomical anomalies that go beyond a simple perineal skin obstruction [4, 5]. A paradox persists in the management of ARM: prenatal diagnosis remains rare during routine morphological ultrasounds. Third-trimester ultrasound rarely identifies the rectal cul-de-sac in isolation [6]. Consequently, the diagnosis relies solely on a systematic clinical examination in the delivery room. A failure in this initial assessment leads to a delayed diagnosis at the stage of acute intestinal obstruction [7]. ARM rarely occurs in isolation. The scientific literature agrees that 60% to 70% of patients have at least one associated anomaly [8]. The VACTERL syndrome (Vertebral, Anal, Cardiac, Tracheoesophageal, Renal, Limbs) necessitates thorough systematic evaluation from the first hours of life [9]. A cardiac ultrasound, an abdominal-pelvic ultrasound, and X-rays of the spine must be performed prior to any surgical procedure to ensure that no life-threatening abnormalities are overlooked.

Initial management of anal atresia should focus on hydration and abdominal decompression; thereafter, surgical intervention will depend on the type of malformation; a simple, limited anorectal malformation may be treated with primary anoplasty, whereas a high or complex malformation will require an initial diversion colostomy followed by definitive surgical repair 4 to 8 weeks later [1,2]. Immediate postoperative care generally involves pain management and infection prevention. Two weeks after surgery, patients must undergo anal dilation to prevent stenosis [3]. Therefore, parental education is essential for achieving good results.

The success of a ARM procedure is no longer assessed in the operating room, but rather at the age of toilet training. The functional prognosis depends intrinsically on the height of the initial malformation and the quality of the sacrum. Low-lying forms typically have an excellent continence rate but are associated with chronic constipation secondary to megarectum, requiring strict laxative monitoring [10].

Conversely, high forms increase the risk of fecal incontinence due to sphincter incompetence. Nearly 40% of children with severe ARM do not achieve socially acceptable continence by adolescence without a bowel management program (antegrade enemas, sphincter rehabilitation), underscoring the importance of long-term multidisciplinary care involving surgeons, pediatric gastroenterologists, physical therapists, and psychologists [10].

5. Conclusion

This condition is routinely diagnosed in the delivery room. Its severity lies in the presence of associated anomalies. Medical management is part of a multidisciplinary approach that includes medical, surgical, radiological, and genetic care.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare that there are no conflicts of interest related to this work.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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