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RECTOURETHRAL FISTULAS IN BOYS: ANATOMICAL AND TOPOGRAPHICAL FEATURES, CLINICAL COURSE AND DIAGNOSTIC CHALLENGES

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Abstract

Anorectal malformations are among the most complex congenital anomalies encountered in pediatric surgery, with some of their forms being associated with significant diagnostic and surgical difficulties. Rectourethral fistulas occurring in boys represent one of the most clinically challenging variants of anorectal malformations. This pathology is often combined with rectal atresia and, in many cases, is not fully diagnosed during the neonatal period. The aim of the study was to determine the incidence of rectourethral fistulas in boys, to analyze their clinical and anatomical characteristics, diagnostic challenges, and their association with concomitant congenital malformations. The study was conducted based on clinical data from patients treated for anorectal malformations between 2009 and 2019. According to the results obtained, rectourethral fistulas accounted for 20.7% of anorectal malformations, concomitant congenital malformations were identified in 73.1% of patients, and incorrect initial diagnoses were recorded in 78.8% of cases. The findings demonstrated the importance of comprehensive instrumental diagnostic methods in the detection of rectourethral fistulas and confirmed that incorrect initial diagnosis increases the risk of surgical complications.

Keywords

anorectal malformations, rectourethral fistula, rectal atresia, diagnosis, pediatric surgery.

Introduction

Anorectal malformations (ARM) represent a group of congenital developmental anomalies of high clinical and social significance [2,7,13]. According to various epidemiological studies, the incidence of ARM averages 1:4,000–1:5,000 live births [2,7]. In recent years, advances in neonatology and prenatal diagnostic capabilities have facilitated earlier detection of these anomalies; however, complex forms of ARM, particularly variants associated with internal fistulae, continue to pose significant diagnostic challenges [1,4,10]. ARM develops as a result of abnormal differentiation of the cloaca during the 4th–8th weeks of embryogenesis [2,16]. Under normal conditions, the cloaca divides into anterior (urogenital) and posterior (anorectal) compartments; disruption of this process leads to pathological communication between the rectum and the urinary tract [2,7]. This embryological mechanism explains the formation of rectourethral fistulae in male patients [5,14,21]. Sex-related differences play an important role in shaping the clinical spectrum of ARM [2,7]. In boys, rectourethral and rectovesical fistulae are more commonly observed, whereas rectovaginal and rectoperineal fistulae predominate in girls [2,11]. These differences are explained by anatomical features of the pelvic organs, characteristics of urogenital system development, and variations in embryological partitioning processes [7,13]. In boys, the prostate and bulbar urethra represent the most frequent sites of rectourethral fistula localisation [5,9,14]. Although rectourethral fistulae are relatively uncommon within the overall structure of ARM, their clinical significance is extremely high [4,8]. This pathology often has a concealed course; pathognomonic signs are not always evident during the neonatal period, which may lead to diagnostic errors and inappropriate surgical management [1,12,18]. Despite this, the literature continues to report persistent difficulties associated with early and accurate diagnosis of rectourethral fistulae [1,4,8].

Materials and Methods. A stepwise diagnostic algorithm was applied in the examination of patients [2,3,10]. At the first stage, the presence of the anus, appearance of the perineal region, pigmentation, depressions, or disruption of the perineal raphe were assessed in

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newborns [3]. Evaluation of the anal reflex allowed indirect assessment of the integrity of muscular structures responsible for the anorectal closure mechanism [10]. At the second stage, instrumental diagnostic methods were employed [5,10,14]. Ultrasonography was used as a primary, non-invasive, and informative modality for assessment of the perineal region and pelvic organs [10]. This method enabled determination of the position of the blind end of the rectum, its height relative to the perineum, and, in some cases, the presence of a fistulous tract [5]. Radiological investigations, including invertography, played an important role in determining rectal height in cases of fistula-free atresia [3,10]. Positioning the patient at a 45° angle ensured optimal distribution of intestinal gas and improved diagnostic accuracy [10]. To identify communication with the genitourinary system, urethrocystography and urethrocystoscopy were considered the principal diagnostic methods [5,12,14]. Antegrade and retrograde irrigography using water-soluble contrast agents allowed evaluation of the distal bowel, as well as determination of the direction and diameter of the fistulous tract [5,14]. Excretory urography was used to assess the functional status of the kidneys and upper urinary tract [11,15]. In complex cases, computed tomography (CT) and magnetic resonance imaging (MRI) were utilised as additional sources of diagnostic information [10,18]. All obtained clinical and instrumental data were compared with intra-operative findings, which increased diagnostic reliability and allowed identification of cases of misinterpretation [4,12].

Results and Discussion. Analysis of the clinical material demonstrated that rectourethral fistulae predominantly occur in combination with rectal atresia. This pathology was identified in 52 patients, of whom 51 had rectourethral fistulae associated with rectal atresia. An H-type rectourethral fistula with a formed anus was extremely rare and was observed in only one case (0.4%). Detection of rectourethral fistulae during the neonatal period was often challenging. In some patients, pathognomonic signs such as meconuria and pneumaturia were observed within the first hours after birth. However, these signs were not present in all cases. Their manifestation depended on the diameter and length of the fistulous tract, as well as the degree of intestinal pneumatisation. In narrow fistulae, temporary obstruction by viscous meconium or mucus was observed, which could mask clinical symptoms. A correct pre-operative diagnosis was established only in a limited number of cases. Rectourethral fistulae were identified intra-operatively in 4 patients (7.7%) who underwent early perineal proctoplasty during the neonatal period. Following perineal proctoplasty, this pathology was detected in 15 patients (28.8%). Among 33 patients who underwent sigmoid colostomy, the pathology was initially assessed as a fistula-free form in 26 cases (78.8%). Comprehensive examinations revealed additional congenital malformations in 73.1% of patients. The most frequent associated anomalies involved spinal pathologies and malformations of the urinary system. The obtained results are consistent with data reported by numerous authors regarding the diagnostic difficulties of rectourethral fistulae [4,8,12]. Several studies emphasise that rectourethral fistulae often occur in association with rectal atresia and have a concealed course during the neonatal period [1,14,18]. The use of comprehensive instrumental diagnostic methods plays a crucial role in reducing diagnostic errors and preventing surgical complications [10,12].

Conclusion

1. Rectourethral fistulae account for 20.7% of anorectal malformations in boys and predominantly occur in association with rectal atresia.
2. Rectourethral fistulae are most frequently localised in the prostatic and bulbar segments of the urethra and are difficult to diagnose during the neonatal period.
3. A high incidence of associated congenital malformations is observed in this pathology, with spinal and urinary system anomalies being predominant.
4. Comprehensive instrumental diagnostics and appropriately selected surgical management improve treatment outcomes and reduce the incidence of iatrogenic complications.

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