H-type anorectal malformation: Case report and review of the literature

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ARTICLE INFO

Article history:
Received 6 January 2014
Received in revised form
30 January 2014
Accepted 3 February 2014
Available online xxx

Key words:
Anorectal malformation
Pediatrics
Congenital malformation
Posterior sagittal anorectoplasty

ABSTRACT

H-type anorectal malformations (ARM) are extremely rare variants in the spectrum of anorectal deformities. This configuration is more commonly described in females, and its presence in males has only been reported in case reports or small series. The report focuses on the successful treatment of this rare anomaly in a male patient and provides a review of currently available literature regarding surgical treatment options and outcomes.

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H-type anorectal malformations (ARM) are extremely rare anomalies with an estimated incidence of 3% of all ARM [1]. In available reports, this variant appears to be found more commonly in Asia (12% of ARM) and in female patients [1,2]. Associated anatomic anomalies are common in these patients, with a potentially higher rate in males [3]. The H-type configuration in males is distinct from the more commonly described anorectal deformities, as a normal or ectopic anus is seen in addition to a fistula to the urinary system [3]. Herein, we report a case of a male with an H-type rectourethral fistula.

1. Case report

The patient is male infant born at a gestational age of 33 weeks who was transferred to our tertiary care center for respiratory distress from an outlying hospital. The patient was intubated upon arrival to our center. He had passed meconium in the first 24 h of life, but on exam was noted to have a slit-like anus (Fig. 1). Particulate matter was also found in the urine. The remainder of the medical history and physical exam was unremarkable. Imaging workup consisted of a renal ultrasound demonstrating debris and air within the bladder, plain abdominal films revealing multiple vertebral segmentation anomalies involving the thoracic spine and sacrum (Fig. 2), and an echocardiogram showing only a small VSD. A barium enema was performed to further delineate the anatomy, which demonstrated a colourethral fistula, most likely to the posterior urethra (Fig. 3). No evidence of urethral stenosis, hypoplasia, or atresia was noted on a voiding cystourethrogram, which did confirm the presence of the fistula as well as right-sided grade II vesicoureteral reflux (Fig. 4).

An exam under anesthesia and divided colostomy was performed. The anal opening was patent but admitted only a 9 mm Hegar dilator without resistance, which was concerning for anal stenosis. A post-operative MRI was obtained, which excluded the presence of a presacral mass, and identified the fistulous tract extending from the anterior wall of the rectum to the prostatic urethra at the level of the levator ani, above the external anal sphincter (Fig. 5). A spinal ultrasound was done after the MRI to better characterize a cystic structure in the region of the filum at the level of the sacral canal that was concerning for a filar or perineural cyst. The ultrasound confirmed the anechoic structure to be a perineural cyst. A follow-up MRI done 6 months later demonstrated complete resolution of such cyst.

At two months of age, the patient underwent posterior sagittal anorectoplasty (PSARP) with ligation of the H-type rectourethral fistula (Fig. 6). The fistula was identified on the anterior wall of the rectum and was quite large and patulous. Post-operatively, a distal
colostogram was obtained without evidence of a remaining or recurrent fistula or stricture. Four months later, the colostomy was closed. The patient currently has regular bowel movements with normal urination six months after the PSARP.

2. Discussion

H-type anorectal malformations (ARM) are extremely rare anomalies in male infants [3]. The embryologic etiology of the malformation is speculative. Stephens et al. have proposed that a misalignment of the cranial and caudal septal components between the genitourinary and gastrointestinal tracts causes these H-type variants [4]. Others attribute the malformation to a persistent cloacal duct [2,5]. Since the defect results from an abnormality in septation, associated anatomic anomalies in the VACTERL spectrum are frequently seen. A series by Rintala et al., which included females, reported major anomalies in 60% of the patients [3]. In order to determine the characteristics and outcomes of this small patient population, we performed a
comprehensive review of the literature to identify similar reported cases of males with an H-type fistula and a normal or ectopic anus (Table 1). Of these cases, only 3/13 patients did not have an associated abnormality. While a normal anus is often encountered, anal or rectal stenosis can be detected in some infants, with one series reporting a 38% incidence [9,10].

The diagnosis of an H-type fistula differs from other ARM patients as a normal anus can be seen on initial examination, thus leading to a delay in diagnosis. Patients can present in childhood with a variety of symptoms including stool in the vestibule in females or in the urine in boys, or recurrent perineal infections. Diagnosis can be made with a combination of contrast studies, MRI, endoscopy, and exam under anesthesia [10]. The use of Hegar dilators during an exam under anesthesia can evaluate for the presence of anorectal stenosis. In a series of patients with H-type malformations in males, misdiagnosis occurred in 2/7 patients [2].

Table 1

<table>
<thead>
<tr>
<th>Study</th>
<th>Age at operation</th>
<th>Normal or ectopic anus</th>
<th>Associated anomalies</th>
<th>Surgical approach</th>
<th>Surgical complications</th>
<th>Functional outcomes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sharma (2002) [6]</td>
<td>3Y</td>
<td>Normal</td>
<td>Spina bifida, sacral agenesis, absent left kidney</td>
<td>Anterior perineal</td>
<td>None</td>
<td>Normal stool/urine at 3 mos post-op</td>
</tr>
<tr>
<td>Stephens (1977) [4]</td>
<td>5Y</td>
<td>Normal</td>
<td>TEF</td>
<td>Anterior perineal</td>
<td>None</td>
<td>Normal stool/urine</td>
</tr>
<tr>
<td>Hong (1992) [7]</td>
<td>5Y</td>
<td>Normal</td>
<td>None</td>
<td>Anterior perineal</td>
<td>None</td>
<td>Normal stool/urine at 15 mos post-op</td>
</tr>
<tr>
<td>Hong (1992) [7]</td>
<td>4Y</td>
<td>Normal</td>
<td>Absent kidney</td>
<td>Anterior perineal</td>
<td>None</td>
<td>Normal stool/urine at 9 mos post-op</td>
</tr>
<tr>
<td>Al-Bassam (1998) [8]</td>
<td>5Y</td>
<td>Ectopic</td>
<td>Pulmonary valve stenosis</td>
<td>Anterior perineal</td>
<td>None</td>
<td>Normal stool/urine at 8 mos post-op</td>
</tr>
<tr>
<td>Rintala (1996) [3]</td>
<td>1 neonatal, 3 childhood</td>
<td>1 normal, 3 ectopic</td>
<td>Vertebral/sacral, renal, cardiac, TEF, malrotation, limb anomalies</td>
<td>2 PSARP, 2 Anterior perineal</td>
<td>Recurrent fistula x 2</td>
<td>Unknown</td>
</tr>
<tr>
<td>Banu (2009) [2]</td>
<td>3D</td>
<td>Ectopic</td>
<td>None</td>
<td>Anterior sagittal anoplasty</td>
<td>None</td>
<td>Normal stool/urine</td>
</tr>
<tr>
<td>Banu (2009) [2]</td>
<td>2D</td>
<td>Ectopic</td>
<td>Cardiac anomaly, sacral hypoplasia, congenital cataract</td>
<td>Anterior sagittal anorectoplasty</td>
<td>None</td>
<td>Died POD 4 (multiple anomalies)</td>
</tr>
<tr>
<td>Banu (2009) [2]</td>
<td>2D</td>
<td>Ectopic</td>
<td>None</td>
<td>Anterior sagittal anorectoplasty</td>
<td>None</td>
<td>Normal stool/urine</td>
</tr>
</tbody>
</table>
Additionally, of male patients with normal or ectopic anal openings who underwent repair of the fistula, 8/13 were not repaired until after the infant period. Thus, either a high index of suspicion of an ARM in the face of other congenital anomalies or an elicited history of urinary symptoms is needed to prompt diagnostic work-up.

There is no consistently recommended surgical approach to repair an H-type ARM [3,10]. Proposed operative approaches have ranged from a simple perineal repair, anterior perineal anorectoplasty, vestibuloanal pull-through, to a posterior sagittal anorectoplasty. The use of a diverting colostomy either initially or with fistula closure has not reached a consensus, as it has not been shown to prevent recurrence [3]. In our own patient, a PSARP was chosen for repair in order to afford adequate exposure of the fistula. If the anatomy is clearly delineated with a contrast study preoperatively, an anterior perineal procedure might be appropriate to decrease the risk of wound dehiscence or local wound complications.

The complications reported after repair include recurrence and wound dehiscence [3,10]. Recurrence rates appear to be lower when the two suture lines (anterior rectum and urethra or vagina) are separate [10]. In the reported H-type cases, all patients had good functional outcomes with both stool and urine, which is not necessarily true for all ARM patients [11]. This may be related to relatively normal anal sphincters and anuses in these patients. In our case, the infant has not yet reached an age for toilet training, so we are unable to reach any conclusions regarding his long term functional outcomes.

3. Conclusion

Based on our review of the literature, infant males with H-type ARM may be more likely than the overall ARM population to have associated anomalies. Despite this pronounced correlation with the VACTERL abnormalities, diagnosis is often delayed. Multiple operative approaches have been reported, and all appear to confer excellent functional outcomes, which may be improved over the more typical anorectal disease.

Disclosures

The authors have no financial support or disclosures to report with respect to the preparation of this manuscript.

References