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Vaginal anomalies and atresia associated with imperforate anus: Diagnosis and surgical management

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ABSTRACT

Background: The association of vaginal atresia (or Mayer–Rokitansky–Kuster–Hauser Syndrome) with imperforate anus is rare and can present significant diagnostic and therapeutic challenges. This study describes clinical characteristics, surgical treatment and outcomes in this group of complex children.
Methods: Records of 20 patients were retrospectively analyzed from two pediatric surgical centers.
Results: Five patients were excluded from the long-term analysis due to inadequate information, leaving long-term follow-up in 15 patients. Mean follow-up was 10 years (range 1–31.1 years). The diagnosis of vaginal atresia was made pre-operatively in 12 out of 15 patients, and in three patients it was identified during the anoplasty. The anorectal malformations were rectoperineal (N = 2), rectovestibular (N = 6), recto-bladder neck (N = 1) and imperforate anus without fistula (N = 6). Satisfactory surgical repair was performed in 13 patients, while one continues to stool through a low perineal fistula awaiting definitive surgery and another underwent a colostomy and mucous fistula. Delayed vaginal reconstruction was due to a failure to identify the problem prior to anoplasty (N = 3). Long-term results demonstrated that anorectal continence was much worse than initially appreciated, and many had associated urinary incontinence. Overall stooling score was far lower than in a separate group of children with imperforate anus without vaginal atresia (Levitt and Peña, 2007).
Conclusions: Vaginal atresia with imperforate anus is a rare and an extensive pre-operative workup of females with imperforate anus must include assessment of vagina patency. Vaginal reconstruction and anorectal continuity can be performed in a variety of approaches, but long-term continence is often not optimal. We propose a pathway for management of this difficult genito-anorectal disorder.

Anorectal malformations affect approximately 1 in 5000 live births [1]. While genitourinary defects can occur in up to 50% of patients with an imperforate anus, vaginal atresia, or Mayer–Rokitansky–Kuster–Hauser Syndrome (MRKHS) associated with imperforate anus is an extremely uncommon surgical problem. The overall incidence of MRKHS is approximately 1 in 5000 female births [2]. This association can present in a delayed manner causing considerable morbidity and distress for the female patient. The combination of both of these anomalies, however, is strikingly rare [3]. While the early identification of imperforate anus is common, an atretic vaginal canal may not be diagnosed until the patient begins menarche with resultant hematocolpos and pain. Early identification of the genital malformation and associated Müllerian duct abnormalities may facilitate the surgical approach. Associated fertility problems emphasize the need for long-term patient and family counseling.

Unfortunately, the predominant literature on this topic comprises only case-report forms [4–9]. One recent large series that describes 7 children with this disorder, as well as several of the patients which are included in another large series of anorectal atresia patients with vaginal anomalies, has advocated for a combined surgical approach [10,3]. The current report presents the largest series to date of patients with MRKHS and imperforate anus. We report the surgical and functional outcomes of these children. As well, we discuss the proper diagnosis and timing of treatment.

1. Methods

This was a retrospective review of 20 patients from two major pediatric surgical centers: C.S. Mott Children's Hospital (United States) and Juntendo University Hospital (Japan). The review was conducted using hospital charts from 1976 to 2013. This study was fully approved by the institutional review boards of both hospitals. Patient characteristics that were abstracted included: level of imperforate anus, associated anomalies, pre-operative workup including radiographic and invasive studies. Surgical procedures were recorded and categorized. Where available, patient's fecal and urinary continence after the definitive surgical therapy was abstracted. We utilized a modification of a previous stooling score to grade overall degree of fecal continence (Table 1) [11,12]. The stooling score is a composite that measures frequency, consistency, odor, continence, soiling,

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sense of urgency and fullness of evacuation. The score ranges from 0 to 36 (with the lower numbers indicating better functional outcomes). The stooling score was stratified into the following groups A (0–12: Good), B (13–23: Fair), and C (24–36: Poor). The score was modified from the stooling score previously used by Hashish et al. [13]. Urinary continence was measured as a yes/no for daytime/nighttime (when age appropriate, scoring only given for children over 3 years of age).

2. Results

2.1. Demographics

A total of 20 children were identified with a combination of these two anomalies (Fig. 1). Review of many of these records dated back to the 1970s, and sufficient data and follow up were not fully present for 5 of these patients. Thus, 15 patients were reviewed in our study; 10 children were from the US and 5 from Japan. The patient demographics are shown in Table 2. As shown in the Table 2, in the majority of cases, anoplasty is approached somewhat later than for most children with imperforate anus. Importantly, the diagnosis of vaginal agenesis was determined preoperatively in most children. One patient in the Japanese series was transferred from an outside hospital amidst a pull-through procedure when the vaginal atresia was identified intraoperatively. Upon transfer, both the imperforate anus and vaginal atresia were corrected. Another patient in the Japanese series underwent a primary anoplasty at an outside hospital; at which time the vaginal atresia was recognized and repaired at a later date. One infant in the US series had the diagnosis recognized only at the time of the anoplasty procedure. In this latter case, only the anoplasty was performed and the vaginal atresia was not addressed surgically.

2.2. Categorization of imperforate anus

Anorectal malformations were classified as rectoperineal (N = 3), rectovestibular (N = 5), recto-bladder neck (N = 1) and imperforate anus without fistula (N = 6).

Table 1
Modified stooling score.

1	**Frequency of defecation:**	*Only w/ enema*	4	**7**	**Time of soiling:**	*None*	0
		Only with suppository	3			*At night*	1
		Every 3 days or more but spontaneously	2			*Both day and night*	2
		Every 1–2 days	1	**8**	**Sense of fullness and evacuation after defecation:**	*Fullness and full evacuation*	0
		Normal (1–2/day)	0			*Fullness but partial evacuation*	1
		Often (3–5/day)	1			*Absent sense of fullness*	2
		6–7/day	2	**9**	**Loss of stool during coughing or crying:**	*No*	0
		8 or more movements/day	3			*Gas*	1
2	**Stool consistency:**	*Hard*	1			*Liquid*	2
		Normal	0			*Solid*	3
		Loose	1	**10**	**Need for medical therapy to control stooling:**	*No*	0
		Liquid	2			*Long period but finally weaned off*	1
3	**Stool odor:**	*Normal odor*	0			*Occasionally*	2
		Offensive odor	1			*Always*	3
4	**Is he/she fully continent?**	*Yes*	0	**11**	**Distension:**	*No*	0
		Partially (occasional accidents)	1			*Mild*	1
		Not continent	3			*Moderate*	2
5	**Requires diapers:**	*None*	0			*Severe*	2
		Night or activity	1	**12**	**For how long since they have been on medications post-op?**	*Less than one month*	0
		Continuously	3			*1–18 months*	1
6	**Soiling:**	*None*	0			*More than 18 months*	2
		Occasional (1–3 times/day)	1				
		Often (4–6/day)	2				
		Permanent (more than 6 times/day)	3				
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		Only with suppository	3			*At night*	1
		Every 3 days or more but spontaneously	2			*Both day and night*	2
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		Normal (1–2/day)	0			*Fullness but partial evacuation*	1
		Often (3–5/day)	1			*Absent sense of fullness*	2
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		Continuously	3			*1–18 months*	1
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		Occasional (1–3 times/day)	1				
		Often (4–6/day)	2				
		Permanent (more than 6 times/day)	3				