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Experiences of Rectovaginal Fistula in Anorectal Malformation☆,☆☆

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ABSTRACT

Background: Rectovaginal fistulas (RVFs) are very rare malformations in females with anorectal malformations (ARMs). Here, we share the clinical features of RVF and report the long-term outcomes.
Methods: RVF patients were classified using a retrospective analysis of ARM patients who underwent operations at Seoul National University Hospital between January 1999 and May 2017. The Krickenbeck continence scoring system was used to evaluate bowel function 5 and 10 years after surgery.
Results: Of the total 460 ARM patients, 203 were female, 7 of whom were diagnosed with RVF. The median age and weight at the time of anorectoplasty were 292 days (range, 140–617) and 8.2 kg (range, 5.5–12), respectively. Six patients had associated anomalies and three patients underwent redo-anorectoplasty. Voluntary bowel movements were observed in 6 out of 7 patients at 5 and 10 years of age. Soiling was observed in all patients at the age of five years and in 6 out of 7 patients at the age of ten years. Constipation was observed in 6 out of 7 patients at both five and ten years of age.
Conclusions: An RVF is a very rare malformation, accounting for 1.5% of total ARMs and 3.4% of ARMs in females. Long-term counseling, education, and guidance are needed for effective management of patients' bowel movements.
Type of study: Prognosis study
Level of evidence: Level IV

Anorectal malformations (ARMs) are a relatively common congenital anomaly. The average incidence is 1 in 4000–5000 live births worldwide, being slightly higher in males [1,2]. Approximately 50%–60% of ARM patients have one or more associated congenital anomalies [3]. In males, the most common type of ARM is the rectourethral fistula, followed by the perineal fistula [4]. In females, the most common type of ARM is the vestibular fistula, followed by the perineal fistula and the cloacal malformation [5,6].

A rectovaginal fistula (RVF) is rare among female ARMs and its occurrence rate is highly variable between reports (0%–84%) [5,6]. In 2002, Rosen et al. published a study of 617 girls with ARMs, reporting 6 RVF patients [6]. Based on this report, the true incidence of RVF is thought to be less than 1%. The reason for the high false incidence of RVF is presumed to be the lack of experience and understanding of RVF, and classification of vestibular fistula or cloacal malformation as RVF. There are also few reports of the long-term outcome for patients with RVF, owing to the scarcity of the condition.

The purpose of this study is to share the clinical features of RVF and to report the long-term outcomes observed at a single center.

1. Methods

We retrospectively reviewed 460 patients who underwent an anorectoplasty for ARM between January 1999 and May 2017 at the Seoul National University Children's Hospital. We classified these patients using the Krickenbeck classification, based on physical examinations, imaging studies and, ultimately, surgical findings [7]. Since March 1998, our institute has applied a protocol for urinalysis, infantography, echocardiography, abdominal ultrasonography, spinal ultrasonography or magnetic resonance imaging, and a detailed physical examination to find associated anomalies for all patients with ARMs.

We investigated gestational age, birth weight, associated anomalies, imaging study results, days of age, and body weight at the time of

Abbreviations: RVF, rectovaginal fistula; ARM, anorectal malformation; VBM, voluntary bowel movement,; PSARP, posterior sagittal anorectalplasty.

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Table 1
Incidence of the various anorectal malformations, as classified by the Krickenbeck classification.

Type	Male / Female	Numbers
Perineal fistula	102 / 55	157
Rectobulbous urethral fistula	70 / 0	70
Rectoprostatic urethral fistula	53 / 0	53
Rectobladder neck fistula	20 / 0	20
Vestibular fistula	0 /92	92
Cloacal malformation	0 / 33	33
Rectovaginal fistula	0 / 7	7
Without fistula	8 / 11	19
Rectal stenosis / atresia	3 / 2	5
H fistula	1 / 3	4
Total	257 / 203	460

surgery, as well as the surgery methodology. Additionally, we analyzed stool frequency, voluntary bowel movements (VBMs), fecal soiling, constipation, and urinary incontinence as postoperative long-term outcomes. These factors were investigated at 5 and 10 years of age. We analyzed the severity of soiling and constipation using the Krickenbeck continence scoring system [7]. We also investigated height and weight at the last outpatient visit. In our study, the median age at the last outpatient visit was 11.8 years (range, 10.1–15.8).

This study was approved by the Institutional Review Board of Seoul National University Hospital (IRB File No. 1807-076-958).

2. Results

The study included 460 ARM patients. Seven patients (7/460, 1.5%) were identified as having RVFs. Of the 203 female patients, vestibular fistulas were the most common (45.3%) type of female ARM, followed by perineal fistulas (27.1%). Among girls, RVF was the fifth most common type of ARM, accounting for 3.4% of all female patients (Table 1).

Six out of 7 patients with RVFs had an associated anomaly. The most common associated anomalies were in the heart and genitourinary system, which were confirmed in 3 patients (Table 2). Among RVFs in our study, there were no spinal anomalies, including sacral deformity or tethered cord. One of the RVF patients was diagnosed with VACTERL association.

The median gestational age and birth weight for the 7 patients diagnosed with RVFs was 40 weeks (range, 38–41.6) and 3.14 kg (range, 2.6–3.45), respectively. All patients underwent a loopogram, and four underwent a cystoscopic examination (Fig. 1). The median sacral ratio of patients was 0.787 (range, 0.748–0.89). All patients underwent a 3-stage operation (stoma formation, corrective surgery, and stoma takedown). Six patients underwent a posterior sagittal anorectoplasty (PSARP), and patient C underwent an abdomino-PSARP owing to the fistula being located near the posterior fornix of the vagina. The fistula was located in the upper vagina in 2 patients, in the middle vagina in 3 patients, and in the lower vagina in 2 patients. The median age and weight at the time of anorectoplasty were 292 days (range, 140–617) and 8.2 kg (range, 5.5–12), respectively. Three patients underwent redo anorectoplasty (Table 3).

Table 2
Associated anomalies in patients with rectovaginal fistula.

Type	Details	Numbers
Heart	VSD (2) ASD (1) PS (1)	3
Gastrointestinal	EA type C (1) Total colonic hypoganglionosis (1)	2
Brain	Small cyst in right caudothalamic area (1)	1
Genitourinary	Double vagina and uterine didelphys (1) Vaginal septum (1) Renal agenesis, Right (1)	3
Musculoskeletal	Clinodactyly, 4th and 5th toes, Right (1)	1

VSD; ventricular septal defect, ASD; atrial septal defect, PS; pulmonary valve stenosis, EA; esophageal atresia.

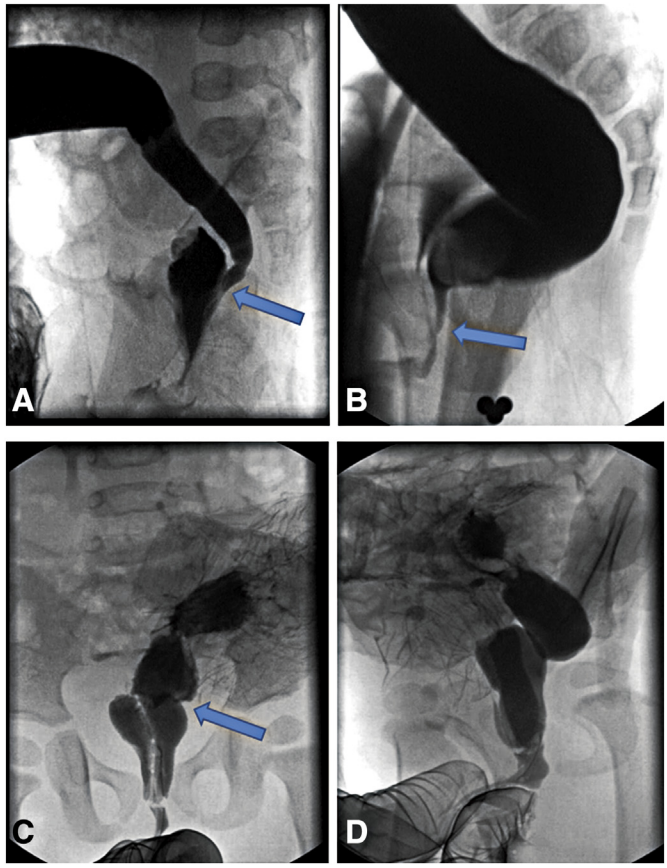

Fig. 1. Contrast study of patients. Figures A and B are distal loopogram of patient A and B, respectively. The patient's rectum did not move to the normal anal position. The contrast medium in the rectum was opacified by vagina through the fistula. Figures C and D are retrograde genitograms of patient D. There were a double vagina and a rectum connected to the left side vagina.

With regards to bowel function, the median stool frequency at 5 and 10 years of age was 1.5 times (range, 0.2–3) and 1 time (range, 0.3–5) per day, respectively. VBMs were seen in 6 out of 7 patients at 5 and 10 years of age. Patient G did not have VBMs at 5 or at 10 years of age, but she was observed to have VBMs upon follow-up at 13 years of age. Fecal soiling occurred in all patients at 5 years of age and, in 6 out of 7 patients, at 10 years of age. The degree of fecal soiling seemed to improve at age 10 compared to age 5. Constipation was observed in 6 out of 7 patients at 5 and 10 years of age, and the number of patients with constipation above grade 2 was higher at age 10 than at age 5 (Table 4). There was no urinary incontinence observed at 5 and 10 years of age in all patients. The median height and body weight at the patients' last outpatient visit was in the 14.3th percentile (range, 1st–96.6th percentile) and in the 30th percentile (range, 12th–82nd percentile), respectively.

3. Discussion

In embryology, the hindgut refers to the caudal part of the digestive tract, which includes the distal third of the transverse colon to the rectum. An ARM is caused by an abnormal hindgut development. During the 7th week of development, the cloacal membrane is divided, creating the anal opening for the hindgut and the ventral opening for the urogenital sinus, with the perineal body forming between the two. The posterior hindgut is closed with ectoderm and is recanalized after

Table 3
Clinical features and surgical outcomes of rectovaginal fistula patients.

Patient	GA (weeks)	Birth weight (kg)	At diagnosis		At anorectoplasty			Redo-anorectoplasty (age)
			Loopogram	Cystoscopy	Age (day)	Weight (kg)	Procedure	
A	38	2.9	Yes	No	292	5.5	PSARP	Yes (2.6); Recurred fistula
B	40	3.4	Yes	Yes	617	12	PSARP	-
C	38	3	Yes	No	203	7.6	Abdomino-PSARP	Yes (4), Mesentery torsion
D	41.6	3.45	Yes	Yes	338	9	PSARP	-
E	41	3.2	Yes	Yes	140	6	PSARP	-
F	39.3	3.14	Yes	Yes	328	8.6	PSARP	-
G	40	2.6	Yes	No	198	8.2	PSARP	Yes (6.5), Hypoganglionosis

GA; gestational age, PSARP; posterior sagittal anorectoplasty, Hypoganglionosis; total colonic hypoganglionosis.

Table 4
Long-term bowel functional outcome of patients with rectovaginal fistula.

Variables	5 years	10 years
Voluntary bowel movement	6 / 7	6 / 7
Soiling, No	0 / 7	1 / 7
Grade 1	2 / 7	5 / 7
Grade 2	5 / 7	0 / 7
Grade 3	0 / 7	1 / 7
Constipation, No	1 / 7	1 / 7
Grade 1	3 / 7	1 / 7
Grade 2	3 / 7	4 / 7
Grade 3	0 / 7	1 / 7

two weeks [8]. Currently, the pathophysiology of ARMs is thought to be the result of an error during the recanalization in the 9th week of gestation and the abnormal positioning of the anal opening in the cloaca. The types of ARMs are related to the extent of development of the posterior aspect of the cloaca. It is believed that, in females, ARMs differentiate into perineal fistulas, vestibular fistulas, rectovaginal fistulas, and cloacal malformations according to the extent of development of the posterior aspect of the cloaca [8,9].

At the Seoul National University Children's Hospital, the type of female fistula was determined through careful perineal bedside examination or under anesthesia, if necessary. When three orifices were present, the boundary between the vagina and anus was clear, the length of the perineal body was sufficient, and the size and shape of the anus were normal, we classified it as normal. When three orifices were present, but the anus was anteriorly positioned without sufficient perineal body length, it was classified as a perineal fistula. When a rectal fistula was found in the vestibule, it was classified as vestibular fistula. If 1 or 2 orifices were found, the type was determined through a contrast study such as a distal loopogram or a retrograde genitogram. If the distinction was not clear from the contrast study, cystoscopy was performed.

In a study published in 2002, the true incidence of RVFs in females with ARMs was reported to be less than 1% (6 RVF patients in 617 female ARM patients) [6]. Previously, RVFs had been considered a relatively common type of ARM in females. In fact, in the articles and books published in 1952 and 1962, Santulli reported the incidence of RVFs as 65% and 60%, respectively [1,10]. Additionally, in 1967, Swenson et al. reported an incidence of 79% for RVFs [11]. This incidence decreased over time, as Santulli et al. reported an incidence of 25% (118 RVF patients in 481 female ARM patients) in 1971 [12], and a study reported in 1988 an incidence of 19% (183 RVF patients in 951 female ARM patients) [13]. A lack of understanding of the accurate anatomy of the different malformations might explain these high false incidences, as Rosen et al. described that cloacal malformations were considered less common than RVFs [6]. Many of our patients had been referred from other hospitals. When classified at other hospitals by an initial examination, RVF was diagnosed in 17 of the patients included in this study (only 2 inborn patients had fistulas). However, once at our hospital and based on physical examination, imaging studies, and surgical findings, 6 of those patients were diagnosed with cloacal malformation, 3 with vestibular fistulas, and 1 patient had no fistula (Fig. 2). Finally, 7 patients were identified as having RVF.

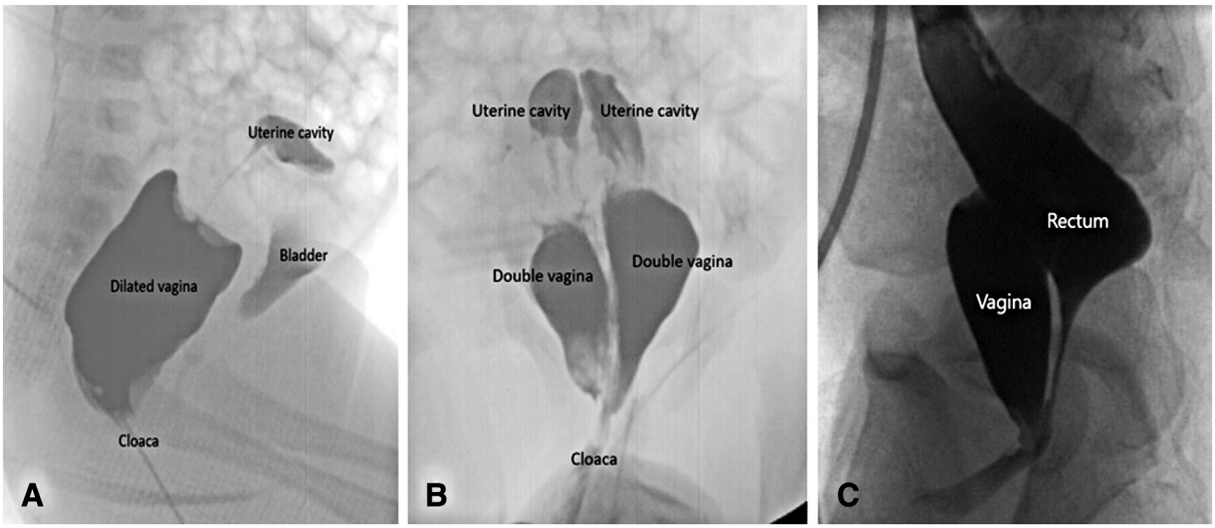

Fig. 2. Contrast study of patients who were incorrectly diagnosed as having rectovaginal fistulas. Figures A and B were retrograde genitograms of the same patient. When injected with contrast medium, double vagina and uterine didelphys were identified, and bladder was simultaneously visualized, so we diagnosed it as cloaca. In this study, communication with rectum was not confirmed. Figure C showed a vestibular fistula in the distal loopogram.

In our study, 3 patients underwent redo operations. Patient A underwent a PSARP and the colostomy was closed 3 months later. After 4 months of stoma closure, recurrence of the rectovaginal fistula was confirmed, and the colostomy was performed again. The patient underwent their first redo-PSARP 3 months later and a stoma closure 6 months afterwards. However, owing to wound disruption from rectovaginal fistula recurrence and infection, the patient underwent another colostomy. The second redo-PSARP took place 5 months later and the patient had a good recovery. Patient C experienced more than 20 episodes of fecal soiling per day after the abdomino-PSARP. Therefore, the patient underwent a redo-PSARP at 4 years of age, during which a 180° torsion of the colonic mesentery was found, and the fecal soiling improved after the correction. Patient G complained of persistent fecal incontinence, and dilatation of the whole colon was found at every examination. Therefore, at 6.5 years of age, after multiple biopsies of the small intestine and colon revealed total colonic hypoganglionosis, the patient underwent a total proctocolectomy with ileal pouch-anal anastomosis.

In a study of long-term bowel function in patients with RVF, Hashish et al. evaluated the bowel habits and quality of life at a mean age of 6.5 years in 48 ARM patients. The mean stooling score was 15.0 ± 3.5 for RVF patients, which was less than the mean stooling score for all ARM patients (16.2 ± 6.2). Additionally, the overall mean quality of life score was 9.0 ± 3.2 for RVF patients, which was less than that of all ARM patients (9.4 ± 3.9) [14]. In their book, Levitt and Peña found that only 50% of RVF patients had VBM, soiling was observed in 75% of patients and only 25% of patients had achieved total fecal continence [15]. In our study, no patients achieved total fecal continence by age 5, and 1 out of 7 patients had achieved it at age 10. In addition, the degree of soiling improved by the age of 10 compared with the age of 5, but constipation was more severe at the age of 10.

Our study reported on RVFs, a very rare type of female ARM. Although our sample size was not large, the value of this report is substantial considering that it addresses a very rare malformation. Our center is a tertiary center in Korea, a hospital visited by patients with rare congenital malformations or diseases from all over the country. For this reason, it is possible that ARMs that required uncomplicated operations were managed in each clinic, and ARMs requiring more complicated treatments were referred to our center. Therefore, the incidence of RVF reported in this study may be overestimated.

4. Conclusion

RVFs are a very rare malformation, accounting for 1.5% of total ARMs and 3.4% of ARMs in females. We found that, in females with ARMs, RVFs could be distinguished from cloacal malformations or vestibular fistulas by accurate physical examination, imaging studies, and via intraoperative findings. Most of the RVF patients had associated anomalies and could be treated with staged operations. Fecal incontinence was observed in all patients at 5 years of age, and in 6 out of 7 RVF patients at 10 years of age. Constipation was also observed in most patients. Long term follow-up of our study showed that most patients had poor bowel function. Therefore, long-term counseling, education, and guidance are needed for effective management of patients' bowel movements.

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