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Case Report

Two Cases of Diverticulitis in Patients With Williams Syndrome

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Williams syndrome is rare and associated with physical anomalies and mental retardation. It is a disease resulting from a gene deletion of chromosome 7. The main concurrent medical conditions typically associated with Williams syndrome are heart defects such as supravalvular aortic stenosis, mental retardation, and unusual physical characteristics. It is also associated with colon diverticulosis and diverticulitis. In the present article, we report on 2 cases of diverticulitis in patients with Williams syndrome, in whom surgery was performed. In many cases of diverticulitis in patients with Williams syndrome, surgical treatment is indicated. It is important to take diverticulitis into consideration when examining a patient with Williams syndrome presenting with abdominal pain and consider surgical treatment if necessary.

Key words: Diverticulitis – Diverticulm – Colon – Perforation – Williams syndrome

Williams syndrome is rare and associated with physical anomalies and mental retardation. It is a disease caused by a deletion of the elastin gene from chromosome 7. The main concurrent medical conditions typically associated with it include heart defects such as supravalvular aortic stenosis, mental retardation, and unusual physical characteristics.[1–5] It may also cause colon diverticulosis and diverticulitis. In this article, we report on 2 cases of diverticulitis in patients with Williams syndrome who underwent surgical treatment for colon diverticulosis.

Case 1

The patient was a 43-year-old man, who underwent surgery for aortostenosis at the age of 15 years and who was at the time infected with hepatitis C through blood transfusion. At 33 years of age, he was diagnosed with Gilbert's syndrome and thereafter was monitored by the Department of Internal Medicine at our hospital. During the course of the follow-up, he was diagnosed with Williams syndrome for unusual facial features and short stature (Fig. 1). At 42 years of age he was hospitalized for

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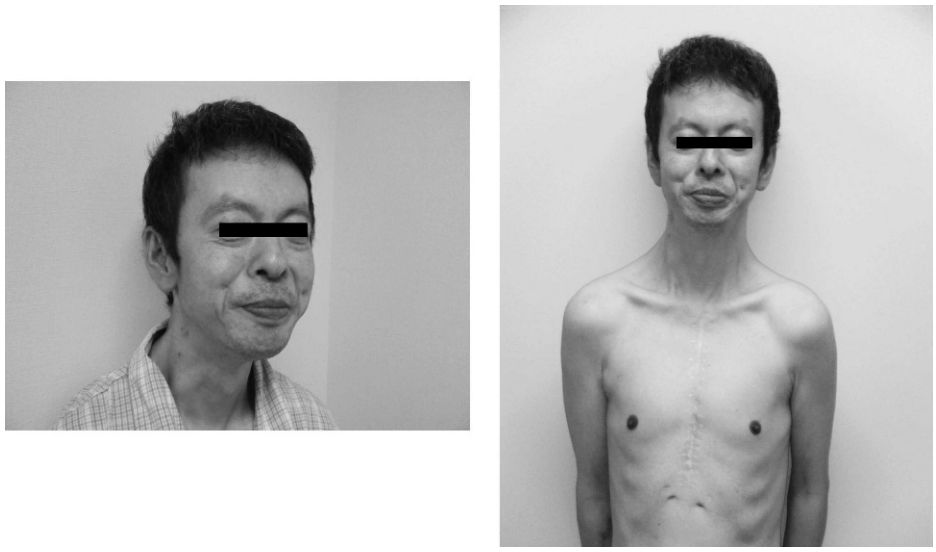

Fig. 1 A 43-year-old man with Williams syndrome: unusual facial features and short stature were observed. A cicatrix from surgery for aortostenosis was observed on the chest.

pneumonia. A barium enema performed subsequently demonstrated multiple diverticula in the sigmoid colon (Fig. 2), whereas the abdominal computed tomography (CT) angiography revealed an intra-abdominal abscess that contained air on the left side of the bladder. The intra-abdominal abscess, having exhibited gradual enlargement, subsided with conservative treatment (Fig. 3). Because no abdominal findings, such as abdominal pain and tenderness, or fever had been observed, conservative treatment was performed. Although abdominal CT images demonstrated an intra-abdominal abscess, there had been no abdominal symptoms, and as there was no abnormality detected by a blood test, the patient had been placed under observation. The abdominal pain then grew worse and symptoms of peritonitis were confirmed. A gastrografin enema revealed multiple diverticula in the sigmoid colon and leakage of gastrografin solution into the extraintestinal space (Fig. 4), and an emergency operation was performed after the diagnosis of perforated peritonitis. A sigmoidectomy and a colostomy were performed. After surgery the patient had no complications and was discharged from the hospital. The pathologic diagnoses determined from surgical specimens were intraperitoneal abscess and perforated sigmoid colon diverticulitis.

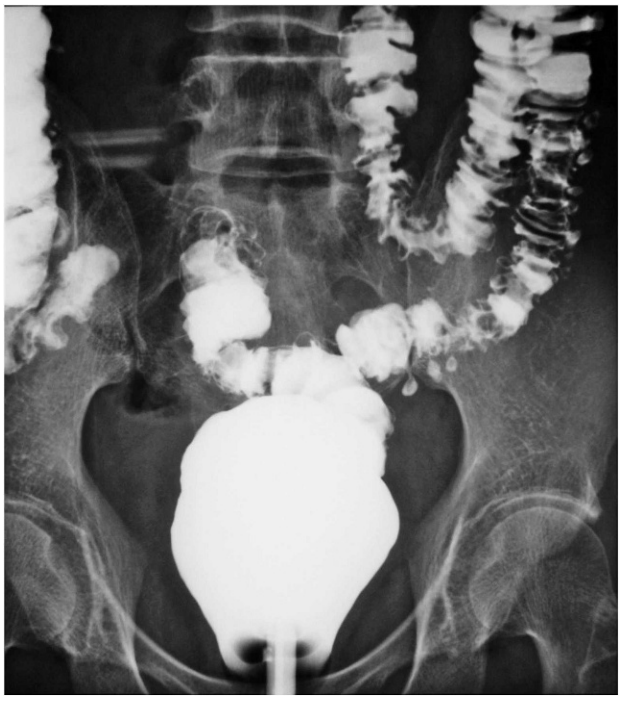

Fig. 2 Barium enema: multiple diverticula in the sigmoid colon were revealed.

Case 2

The patient was a 71-year-old woman who had been diagnosed with Williams syndrome for unusual facial features and short stature. As a result of a careful examination of abdominal pain and anemia, as well as fecal occult blood testing, many diverticula in, and severe stenosis of, the sigmoid colon were found. An endoscopic examination of the colon revealed many diverticula in the sigmoid

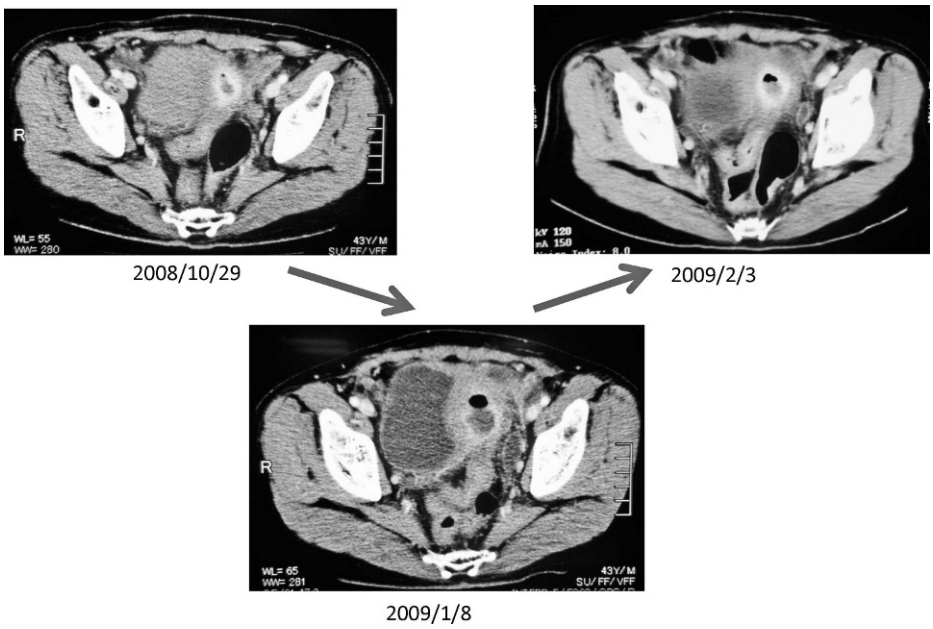

Fig. 3 Abdominal CT angiography: an intra-abdominal abscess that contained air was found on the left side of the bladder, which exhibited gradual enlargement. The abscess subsided with conservative treatment.

colon, some being extremely deep. It became extremely narrow and a colonoscopy could not go through the stenosis. In biopsies from the tissues around the area of the stenotic lesion, no malignant finding was observed (Fig. 5). Thereafter, the patient developed symptoms of peritonitis and underwent surgery for suspected perforated diverticulitis. The left side of the colon was resected and a transverse colostomy was performed. After surgery the patient had no complications, recovered well, and was discharged from hospital. The pathologic diagnosis determined from surgical specimens was diverticulitis of the sigmoid colon with an abscess.

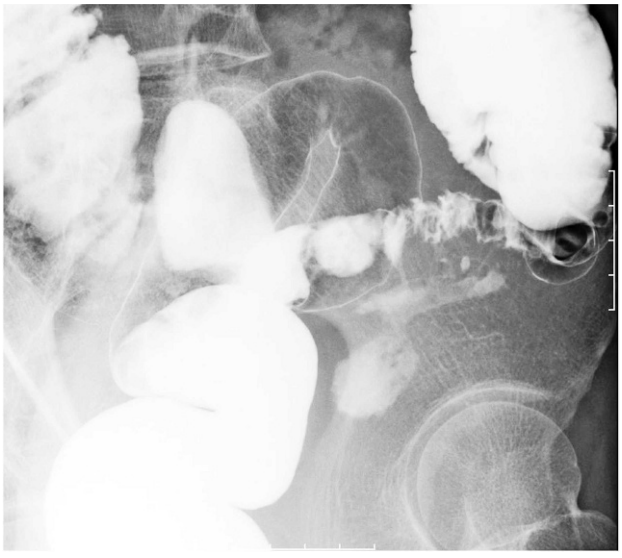

Fig. 4 Gastrografin enema: multiple diverticula in the sigmoid colon and leakage into the extraintestinal space were observed.

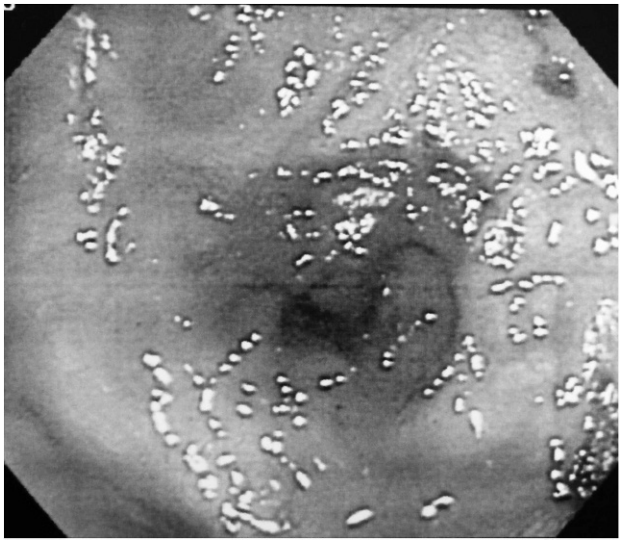

Fig. 5 Endoscopic examination of colon: many diverticula in the sigmoid colon were found, some being extremely deep. It became extremely narrow and a colonoscopy could hardly go through the stenosis. In biopsies from the tissues around the area of the stenotic lesion, no malignant findings were observed.

Table 1 Patient characteristics, findings, and treatment

Patient no.	Gender	Age at diagnosis (y)	Findings	Treatment
1	Male	18	Appendicitis, abscess, sigmoid diverticulitis, adhesion ileus	Sigmoid resection, adhesiolysis, appendectomy
2	Male	17	Appendicitis, perforated sigmoid diverticulitis. relapse of diverticulitis with fistula	Appendectomy, temporary colostomy, sigmoid resection, anus praeter and reanastomosis
3	Male	19	Perforated sigmoid diverticulitis, peritonitis, scar herniation	Sigmoid resection, anus praeter, reanastomosis, abdominal wall reconstruction
4	Male	23	Sigmoid diverticulitis, perforation, peritonitis	Sigmoid resection, anus praeter
5	Male	24	Sigmoid diverticulitis, perforation, peritonitis, abdominal abscess	Sigmoid resection, anus praeter, reanastomosis
6	Male	26	Sigmoid diverticulitis, perforation, peritonitis	Sigmoid resection
7	Male	31	Sigmoid diverticulitis	Sigmoid resection, adhesiolysis, appendectomy
8	Male	35	Sigmoid diverticulitis (rectal prolapse 2 y earlier)	Conservative treatment
9	Female	40	Sigmoid diverticulitis, perforation, peritonitis	Sigmoid resection
10	Female	36	Sigmoid diverticulitis, appendicitis	Appendectomy, conservative treatment
11	Male	17	Sigmoid diverticulitis, perforation, abdominal abscess	Sigmoidectomy with a Hartmann's procedure
12	Male	42	Uncertain	Uncertain

Discussion

Williams syndrome was first reported by Williams and Beuren in 1961–1962 and its common symptoms are unusual facial features, heart defects (mainly supravalvular aortic stenosis), growth retardation, mental retardation, specific cognitive impairment, and hypercalcemia.[1–3] It is a rare disease, with an estimated prevalence of 1 in 20,000 births, and a possible cause of the disease deletions of the elastin gene from chromosome 7 (at 7q11.23) have been reported.[3–5] Colon diverticulosis is seen in relatively young patients and diverticulitis often causes serious complications.[6] Diverticulosis has been documented in 18% of patients with Williams syndrome.[7] Diverticulosis is caused by increased pressure in the colon and by changes in elastin and collagen of the colonic wall, which occur with age. It was reported that elastin gene mutations also cause diverticulosis.[8] Another report, however, argued that the disease is caused by ischemia induced by complicated colonic causes.[9] Using the PubMed database, we searched using the keywords ''Williams'' and ''Diverticulitis'' and found 12 adult patients with Williams syndrome who had colon diverticulitis as a complication.[8,10,11] Among these 12 patients, 10 patients underwent surgery, 2 patients recovered with conservative treatment, and 1 patient eventually passed away (Table 1).[8,10,11] It is therefore believed that, in many cases of diverticulitis in adult patients with Williams syndrome, a surgical operation is indicated. In fact, the 2 patients we reported on underwent surgical treatment. It is therefore necessary to take diverticulitis into consideration when examining a patient with Williams syndrome presenting with abdominal pain and consider the indication of surgical treatment.

Acknowledgments

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