Gastric Submucosal Hematoma and Mucosal Rupture with Ehlers-Danlos Syndrome: A Case Report

Yasuo Kobayashi1, Akifumi Kuwabara1, Haruhiko Okamoto1, Kazuhito Sugimura2 and Katsuyoshi Hatakeyama1

1 Division of Digestive and General Surgery; and 2 Divisions of Gastroenterology and Hepatology, Graduate School of Medical and Dental Sciences, Niigata University, Niigata, Japan

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Summary. Ehlers-Danlos syndrome (EDS), a rare disease caused by the inadequate production of connective tissue collagen, can carry a high risk of life-threatening aneurysms or intestinal bleeding. Recently, we experienced a case of EDS with very unusual clinical features. A 20-year-old woman presented at our hospital with a sudden onset of hematemesis. She had been previously diagnosed with EDS but had received no specific treatment for this disorder. Results of a gastrointestinal endoscopy suggested duodenal intussusception. We therefore performed an emergent operation, which revealed a submucosal hematoma at the antrum, the mucosal rupture of which had led to hematemesis. The patient underwent a distal gastrectomy and recovered without any complications after the operation. This case highlights the fact that patients with EDS may present with quite unexpected symptoms or images that may still be related to the underlying pathogenesis of this disorder.

Key words — Ehlers-Danlos syndrome, gastric submucosal hematoma, intestinal bleeding.

INTRODUCTION

Ehlers-Danlos syndrome (EDS), a congenital connective tissue disorder characterized by skin hyperextensibility, joint hypermobility, and vascular fragility, can lead to life-threatening gastrointestinal and vascular complications1-4). Here we present a case of EDS that developed a gastric submucosal hematoma whose mucosa ruptured, mimicking duodenal intussusception, and which was successfully treated by extensive distal gastrectomy.
Fig. 1. Chest-abdominal radiography. a. Scoliosis and b. Elongation of the stomach are observed.

Fig. 2a. Gastrointestinal endoscopy. a. There appears to be a cylindrical mucosal tubercle (about 5 cm long) at the antrum though no pyloric ring appears anywhere. b. Note an ulcer-like indentation at the top of the tubercle.

Fig. 3. Although the computed tomogram shows a target sign-like structure in the antrum (arrows), no a multiple concentric ring sign, or a distinctive feature of intestinal intussusception, can be seen.
treat the duodenal intussusception. However, during the operation it became apparent that she actually had a large submucosal hematoma at the antrum (Fig. 4). We suspect that this hematoma was the cause of hematemesis. The submucosal hemorrhage had spread to cover a considerable area of the stomach, and therefore its distal side was extensively resected. The resected specimen revealed a ruptured circumferential mucosa with a massive hematoma (approximately 160 × 140 mm) (Fig. 5). On histology, the massive hematoma was associated with numerous inflammatory infiltrates and vessels of various sizes in the edematous submucosal layer of the stomach. We reconfirmed the previous CT scan picture; a multiple concentric ring sign, a distinctive feature of intestinal intussusception, was not observed.

Although the respirator tried to collect the air that she expired during the operation, her main bronchus
collapsed and was obstructed, interrupting usual respiration. For this reason, she was kept in the intensive care unit for two days after surgery, during which time her breathing improved. She recovered without any further complications and was discharged on day 15 after the operation.

The patient has not shown any gastrointestinal problems as of January, 2006.

DISCUSSION

The critical complications of EDS are an aortic aneurysm and colon perforation1-3). Vascular fragility means even slight pressure can lead to hematoma, which can produce intestinal bleeding which, although not common, is one of the most critical complications of this disorder4). In the present case of EDS, underlying vascular fragility was likely to have led to form the hematoma that subsequently ruptured.

EDS has been classified into nine types according to clinical and genetic features, and each type arises due to different causes4). The "ecchymotic type IV" has the most clinical significance because it can be accompanied by serious complications5-7), especially in the vascular and digestive systems. Although the current case did not have any apparent abnormalities of the vascular system, and the type of EDS has not yet been determined, it is likely that this disease was type IV EDS accompanied with serious gastrointestinal troubles. There was no evidence of any trigger such as bruising or cough before hematemesis, suggesting that intestinal hematomas in EDS patients may be very fragile and prone to rupture.

EDS cases with complications of vascular rupture reported to date have developed mostly from aneurysms in various arteries8-9), or less commonly from peptic ulcerations, hiatus hernia, colonic diverticula, or an external hemorrhoid9). No cases of an intestinal submucosal hematoma have been reported, however. The present case illustrates that the intestinal bleeding in EDS may result from unusual origins. If a submucosal hematoma arises in the stomach, it may grow considerably in the large gastric space without showing any symptoms such as obstruction or bleeding. In addition, if a hematoma develops at the antrum, it may obscure the narrow orifice of the pyloric ring, which in the present case led us to diagnose initially a duodenal intussusception by gastrointestinal endoscope.

Furthermore, since the symptoms of type IV EDS may be indistinct and atypical4), some cases of type IV EDS may not be diagnosed before operation. Therefore, EDS should be considered in patients with unexpected sudden intestinal hemorrhage as well as in those with unexpected colon perforation.

This case highlights the fact that the patients with EDS may present with quite unexpected symptoms or findings that may still be related to the underlying pathogenesis of this disorder. If a known EDS patient shows hematemesis, a gastrointestinal endoscopy and a computed tomography should be performed immediately.

REFERENCES