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

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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 complications[1-4]. 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.

CASE REPORT

A 20-year-old woman presented at our hospital with a sudden onset of hematemesis on May 21st 2004. According to her medical history, she had been clinically diagnosed with EDS at a younger age but had received no specific treatment for this disorder. Her only noteworthy family history was that her mother suffered from rheumatoid arthritis. On physical examination, she had peculiar features at the extremities and a skeletal structure that suggested hyperextensibility with acroteric atrophy, joint hypermobility, and deformity such as from scoliosis (Fig. 1). Laboratory examination showed mild leukocytosis, anemia, and prolonged prothrombin time (WBC 14200 /μ, Hb level 10.7 g/dl and PT 57 %). She had no pain or tenderness in the abdominal region. A gastrointestinal endoscopy was performed immediately and revealed unusual findings (Fig. 2). There was a cylindrical mucosal tubercle (about 5 cm long) at the antrum, but the pyloric ring could not be found. Closer observation indicated an ulcer-like indentation at the top of the tubercle, which led us to suspect duodenal intussusception. An abdominal CT scan showed a target sign-like structure at the antrum (Fig. 3).

The patient underwent an emergent operation to

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Abbreviations: EDS, Ehlers-Danlos syndrome.

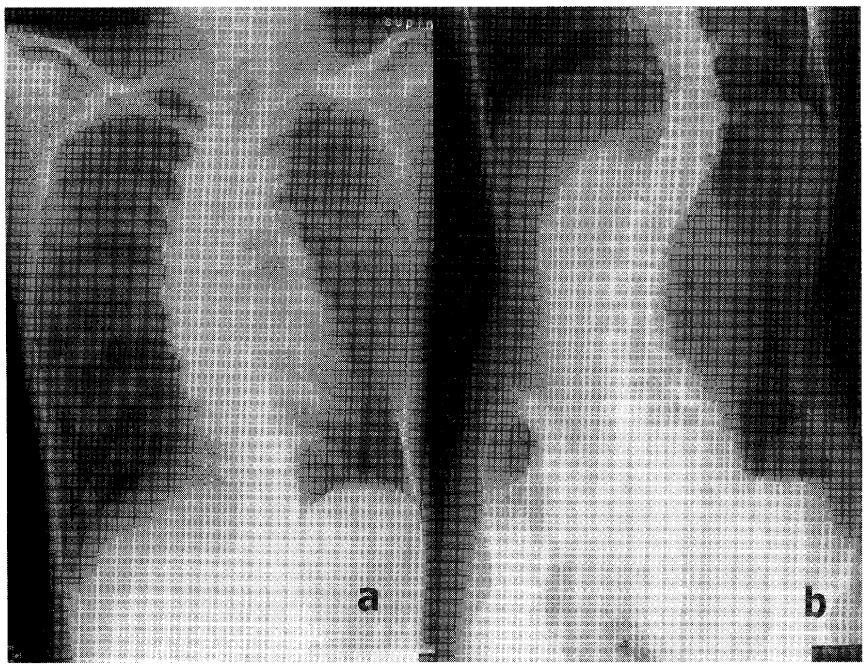

Fig. 1. Chest-abdominal radiography. **a.** Scoliosis and **b.** Elongation of the stomach are observed.

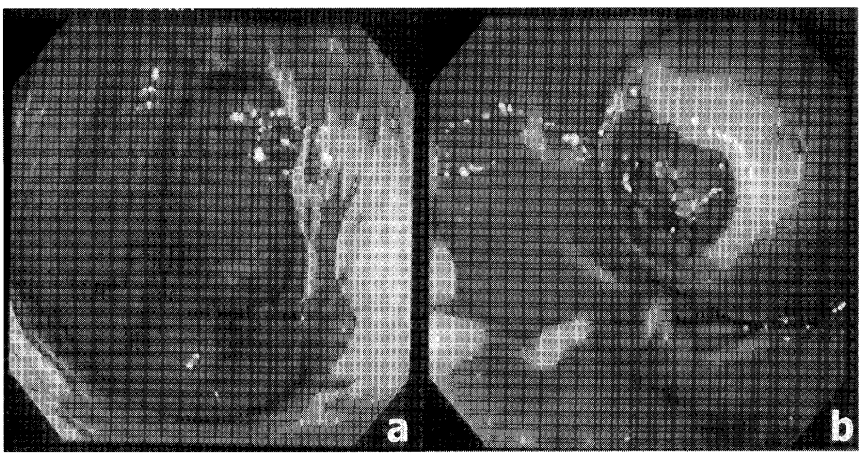

Fig. 2a. Gastrointestinal endoscopy. **a.** There appears to be a cylindrical mucosal tubercle (about 5 cm long) at the antrum though no pyloric ring appears anywhere. **b.** Note an ulcer-like indentation at the top of the tubercle.

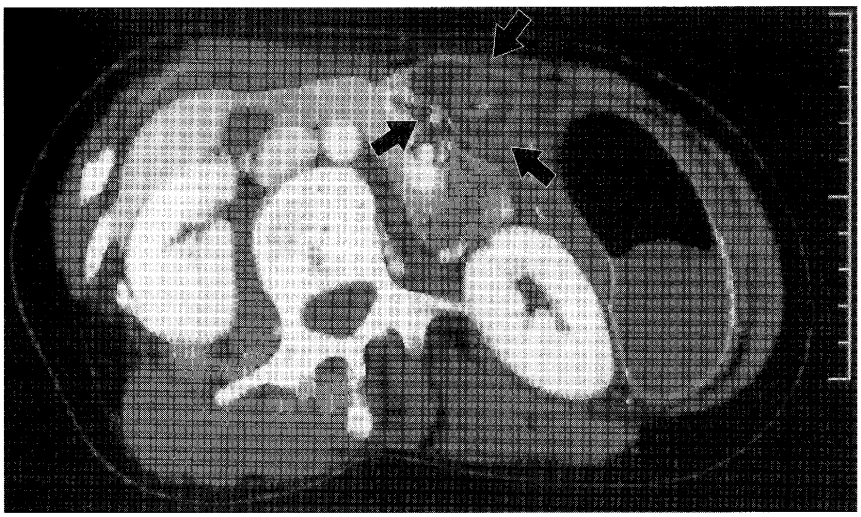

Fig. 3. Although the computed tomogram shows a target sign-like structure in the antrum (*arrows*), no a multiple concentric ring sign, or a distinctive feature of intestinal intussusception, can be seen.

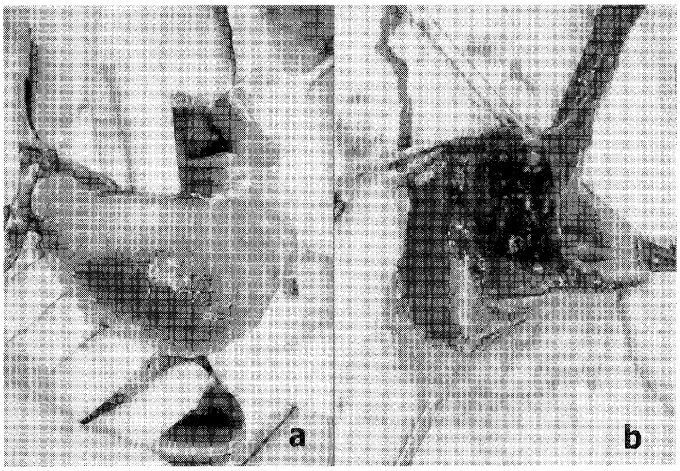

Fig. 4a. During the emergent operation, the submucosal hemorrhage is seen to have spread to a considerable area of stomach from the serous side, and **b.** Observation of the stomach by gastrotomy at the anterior wall of antrum shows a large hematoma in the gastric wall.

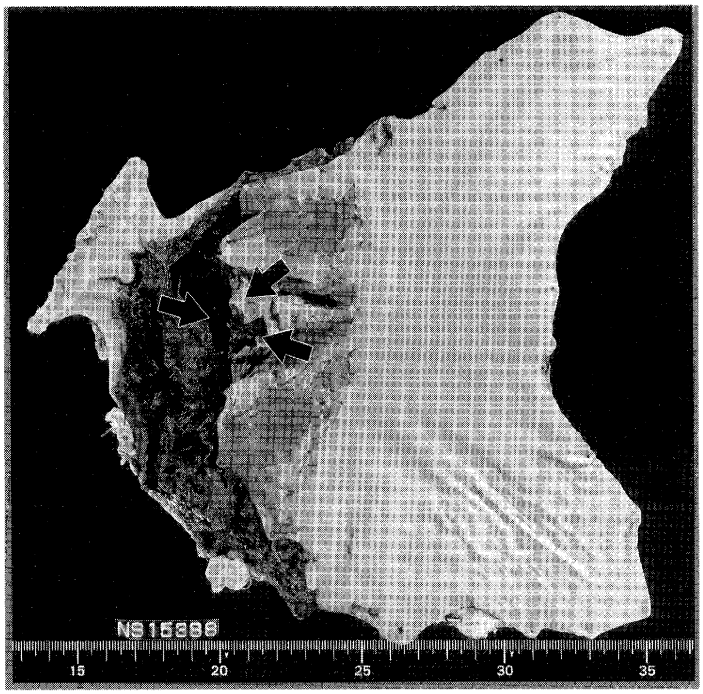

Fig. 5. There is a massive hematoma (approximately 160 × 140 mm) in the stomach. The circumferential mucosa of the antrum has been completely ruptured (*arrows*).

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 perforation[1,3-5]. 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 disorder[6]. 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 causes[4]. The "ecchymotic type IV" has the most clinical significance because it can be accompanied by serious complications[1,5,7], 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 arteries[1,8], or less commonly from peptic ulcerations, hiatus hernia, colonic diverticula, or an external hemorrhoid[9]. 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 atypical[4], 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.

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