Celiac artery disease and fatal rupture of a hepatic artery aneurysm in the Ehlers-Danlos syndrome

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Isolated visceral arteriopathies of the celiac and hepatic artery are rare. We present a case of a Caucasian man who presented with abdominal pain and was found to have a spontaneous celiac artery dissection. Genetic analysis revealed a mutation consistent with Ehlers-Danlos syndrome type IV. The patient died 2 months later from a spontaneous rupture of his hepatic artery.

CASE REPORT

A 31-year-old man with fragile, easily bruisable skin, abnormal (atrophic) scarring, joint hypermobility, and extensive varicosities of the legs presented with sudden onset of left upper quadrant abdominal pain. His blood pressure was 146/75 mm Hg and heart rate, 114 beats/min. The epigastric region was tender and the left upper quadrant of his abdomen was soft. His joints were hypermobile, his skin over the chest was translucent, and severe varicose veins were present on his lower torso and lower extremities. A complete blood count, basic metabolic panel, liver function tests, activated partial thromboplastin time, prothrombin time/international normalized ratio, and amylase and lipase levels were within normal limits (*Table*). An electrocardiogram showed only sinus tachycardia, and an echocardiogram was normal. Computed tomography (CT) of the abdomen revealed high-density fluid around the pancreas and spleen. A serologic workup for a vasculitis was negative.

Due to a normal amylase and lipase level and persistent abdominal pain, a CT angiogram of the thorax and abdomen was performed and revealed a celiac artery dissection with associated thrombus extending into the splenic artery (*Figure*). The spleen was infarcted. The patient was managed with anticoagulation by continuous heparin infusion and metoprolol. He responded well to conservative management and was discharged home. There was a clinical suspicion of Ehlers-Danlos syndrome (EDS) type IV. The patient's paternal grandmother also had extensive varicosities. Molecular studies revealed a mutation in the *COL3A1* gene, which is consistent with the diagnosis of EDS type IV.

Approximately 2 months after hospital discharge, the patient complained once again of sudden onset of severe abdominal

Table. Patient laboratory values

Test	Result
Sodium (mmol/L)	138
Potassium (mmol/L)	3.7
Chloride (mmol/L)	102
Bicarbonate (mmol/L)	28
Blood urea nitrogen (mg/dL)	11
Creatinine (mg/dL)	0.7
Glucose (mg/dL)	116
Alanine aminotransferase (U/L)	27
Aspartate aminotransferase (U/L)	21
Bilirubin total (mg/dL)	0.25
Lipase (U/L)	26
Amylase (U/L)	78
White blood cells (K/ μ L)	15.1
Hematocrit (%)	42.8
Hemoglobin (%)	15.2
Platelet (K/ μ L)	321
International normalized ratio	1.0
Partial thromboplastin time (seconds)	25
Antineutrophil cytoplasmic antibodies	Negative
Antinuclear antibody	Negative

pain. As he and his wife were on their way to the car to drive to the emergency department, he collapsed and died. Autopsy revealed a ruptured hepatic artery aneurysm with 2.5 to 3 L of fresh blood in the peritoneal cavity. Their infant son did not have the *COL3A1* mutation.

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Figure. CT angiogram of the abdomen (sagittal view) showing an isolated celiac artery dissection.

DISCUSSION

Edvard Ehlers (1863–1937) in Denmark and Henri-Alexandre Danlos (1844–1912) in France independently published lucid descriptions of the condition that would later bear both their names; Ehlers published in 1901 and Danlos in 1908 (1). Now many types of EDS are recognized. All three patterns of Mendelian inheritance are represented (autosomal dominant, autosomal recessive, and X-linked recessive) (2). EDS type IV, also known as the vascular type, is a rare connective tissue disorder with autosomal dominant transmission (McKusick catalog number 130050) caused by mutations in the *COL3A1* gene. As a result, these patients have increased fragility of connective tissue with arterial, intestinal, and uterine ruptures. The estimated prevalence for types of EDS varies between 1 in 10,000 and 1 in 25,000, with EDS type IV representing approximately 5% to 10% of cases (3). The median age of death is 50 years (3, 4).

Search of medical publications to 1959 revealed only 39 cases of isolated celiac artery dissections, making it the least common visceral artery dissection reported. Treatment options include surgery, endovascular repair, or medical management (3).

1. Johns Hopkins Medicine. *Online Mendelian Inheritance in Man*. Available at www.omim.org.
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4. Ganesamoni R, Agarwal S, Stephen E, Narayan RL. Isolated celiac artery dissection with splenic infarction: report of a case. *Eur J Vasc Endovasc Surg* 2007;13(4):58–59.