CASE REPORT

Management of Spontaneous Liver Hematoma in Ehlers-Danlos Syndrome Type IV: A Case Report

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ABSTRACT

Introduction: Liver hematoma is an uncommon feature of Ehlers-Danlos syndrome (EDS) type IV. The limited literature that exists to guide management does not establish a standard of care.

Case Presentation: A 26-year-old man presented with acute abdominal pain caused by a large, spontaneous liver hematoma. Invasive prophylactic arterial embolization was done twice, but surgical evacuation was not offered because of concern for poor healing and brittle vasculature, later diagnosed as symptoms of the patient’s EDS type IV. During hospitalization the patient died of spontaneous intracerebral and intra-abdominal hemorrhaging.

Conclusion: This case illustrates a nonsurgical management option for spontaneous liver hematoma in a patient with EDS type IV. An interdisciplinary team should help guide care, including consideration of invasive procedures such as arterial embolization and surgery. Patient and family education, genetic testing, and timely medical record documentation may reduce the morbidity and mortality of patients with this syndrome.

INTRODUCTION

Ehlers-Danlos syndrome (EDS) is a heterogeneous and inherited group of connective tissue disorders. Thirteen phenotypically distinct EDS subtypes exist.1 EDS type IV, or vascular subtype, is autosomal dominant and is associated with spontaneous arterial, uterine, and colonic rupture caused by structurally abnormal type III collagen.1 Liver hematoma is an uncommon feature of EDS type IV, and the limited literature on its management includes descriptions of intraoperative and postoperative deaths.2-4 A standard of care is not established. Exhaustive unsuccessful attempts were made to reach the patient’s next of kin. An effort has been made to anonymize patient information.

CASE PRESENTATION

The institutional review board determined that this case study did not meet the criteria for human subjects research and therefore did not require board approval.

Presenting Concerns

A 26-year-old man was watching TV in bed when he experienced acute, severe abdominal pain. The pain felt similar to when he sustained a spontaneous liver hematoma 7 years earlier, which necessitated a right hepatic lobectomy; thus, he went to the Emergency Department. An abdominal computed tomography (CT) scan revealed a 14-cm subcapsular liver hematoma, so he was admitted to the intensive care unit for hemorrhage monitoring. Concern was raised for EDS because he also had a spontaneous pneumothorax 2 years earlier and a younger teenaged brother had died of a spontaneous aortic rupture. The hepatobiliary surgical team was consulted for hematoma evacuation; however, surgery was not offered because of concern for a poor surgical outcome subsequent to poor healing and the brittle vasculature believed to be caused by EDS.

Therapeutic Intervention and Treatment

During the following 4 days, 3 episodes of tachycardia, worsening pain, and increased intra-abdominal pressure suggested episodes of recurrent bleeding (Figure 1). Results of a repeated CT scan showed enlargement of the hematoma. Results of an angiogram performed by Interventional Radiology showed no extravasation; therefore, the non-bleeding left hepatic artery was prophylactically embolized (Figure 2A). Later, the right hepatic artery branches reconstituting the left hepatic artery were also prophylactically embolized (Figure 2B). Invasive interventions were stopped thereafter because the bleeding was attributed to capillary leakage from capsule shearing. He received medical management of pain and anemia.

Follow-Up and Outcomes

The patient’s bleeding was believed to have stopped once his pain improved. His mentation remained intact, and tachycardia resolved, so he was transferred to a ward. That night, he experienced a hyperacute headache, with systolic blood pressure above 200 mmHg. Evolving neurologic impairment developed during the physical examination. Results of a CT scan revealed a large subarachnoid and intraventricular hemorrhage resulting from a dissecting right posterior inferior cerebellar aneurysm. He was intubated and emergently transferred to the regional neurosurgery center to get an external ventricular drain plus aneurysm clipping.

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He remained intubated and sedated for 3 more days; however, he died on hospital day 14 after an acute intra-abdominal hemorrhage. Genetic test results eventually revealed a \textit{COL3A1} gene variant (c.1610G>T; p.Gly537Val) consistent with EDS type IV. A timeline of the case appears in Figure 3.

**DISCUSSION**

This case illustrates a management option for spontaneous liver hematoma in a patient with EDS type IV, an uncommon complication of a rare disease. Interdisciplinary management by intensivists, hepatobiliary surgery, interventional radiology, neurosurgery, genetic testing, and social work are challenging but beneficial for these medically fragile patients. Clinicians should use symptoms such as pain, signs such as tachycardia and increased intra-abdominal pressure, and imaging findings to identify recurrent bleeding episodes that compel invasive intervention. Prior case reports on EDS-related subcapsular liver hematomas recommend avoiding surgical intervention, so we attempted embolectomy to achieve source control. However, the patient did recover from a previous liver hematoma evacuation and lobectomy, suggesting he may have tolerated another surgery. In the end, the patient may have benefited
Relevant medical history
A 26-year-old man presented with a chief concern of abdominal pain. He had a history of a spontaneous liver hematoma that led to a right hepatic lobectomy at age 19, a spontaneous pneumothorax at age 24, and his brother died as a teenager of a spontaneous aortic rupture.

Physical examination: Tachycardia, acute distress, tender and distended abdomen

Diagnostic evaluations: CT of abdomen, CBC

Diagnoses: Large liver hematoma, anemia

Day 0
Initial treatment included pain control, hepatobiliary surgery and IR consultation, and ICU monitoring for frequent vital sign and abdominal circumference checks.

Day 1
Acute pain and tachycardia lead to another abdomen CT, showing a larger liver hematoma. Results of IR angiogram showed no extravasation, leading to prophylactic left hepatic artery embolization.

Day 2
Patient had acute pain and tachycardia; CT scan was not performed. IR angiogram results showed no extravasation, leading to prophylactic right hepatic artery embolization.

Day 4
Geneticist and social worker were consulted. Genetic testing was ordered. Patient had acute pain and tachycardia; received pain management only.

Day 11
The patient was transferred to ward. Acute headache and neurologic deficits lead to CT of head, showing intracerebral hemorrhage. Patient was transferred to regional neurosurgery center; aneurysm clipped and external ventricular drain placed.

Day 14
Patient had acute tachycardia and abdominal distention caused by intra-abdominal hemorrhage, leading to death.

After patient’s death, genetic test confirmed Ehlers-Danlos syndrome type IV.

Figure 3. Case timeline. CBC = complete blood cell count; CT = computed tomography; ICU = intensive care unit; IR = interventional radiology.
from hematoma evacuation for pain relief and to avoid the fatal large intra-abdominal hemorrhage.

The patient’s family planning may have been affected by an honest discussion with clinicians after his first liver hematoma; at the time of his death, he had a young child, and his wife was pregnant. This time, a genetic consultation was obtained early in the hospitalization. The genetics team oversaw the genetic testing and counseled the patient and family throughout hospitalization. Once the patient’s COL3A1 gene variant was confirmed, outpatient family genetic counseling and testing were done. The patient’s child, nephew, and sister had the same variant. They now all carry the Ehlers-Danlos Society Wallet Card containing emergency information; wear medical alert bracelets; have updated medical records including emergency, colonoscopy, and pregnancy recommendations; and have been given handouts of warning signs and symptoms to keep at home. His child now gets regular echocardiograms and cardiology appointments to monitor for aortic aneurysms. His nephew stopped playing contact sports. His sister has since had a carotid cavernous fistula requiring drainage and embolization; her presenting symptoms were headache and diplopia, and her medical record documentation of EDS type IV led to a quick and accurate diagnosis. Imaging also revealed many healed carotid dissections and pseudoaneurysms, so she will receive aspirin therapy indefinitely to prevent embolic strokes. We encourage clinicians to have honest communication with patients and families about genetic inheritance and the high morbidity and mortality of EDS type IV, even in unconfirmed cases.

CONCLUSION

EDS is a very rare connective tissue disorder that impacts many organ systems. Because of the rarity of this disease, even rarer complications and their management have not been well studied. Patient and family education, genetic testing, and timely medical record documentation may reduce the morbidity and mortality of patients with EDS type IV.

Disclosure Statement

The author(s) have no conflicts of interest to disclose.

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Authors’ Contributions

All authors assisted in study design, data collection, and manuscript preparation. All authors have given final approval to the manuscript.

Prior Presentation

Dr Imp presented the case at the American College of Physicians Northern California Chapter Meeting in Santa Clara, CA, on November 3, 2018. He was a finalist in the case report poster competition.

How to Cite this Article


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