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Inferior Vena Cava Thrombosis in a Porphyria Patient

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INTRODUCTION

This case report discloses an inferior vena cava (IVC) thrombosis event after a cardiac catheterization procedure in a patient with porphyria. As a complication, IVC thrombosis in patients undergoing cardiac catheterization is not common. On the other hand, porphyria is also a rare disease in our population. The relevance between these two could not establish with sporadic related reports and no definite academic interpretation. However, it is a precious experience in attention to the thrombotic events related to invasive intravascular procedures when caring porphyria cases in the future.

CASE REPORT

This female patient was diagnosed with porphyria at the age of 44. The diagnosis took at least 9 months from the insidious onset. The disease was brought to light due to the unexplained intermittent abdominal pain, also some symptoms presenting on and off, including nausea, dizziness, headache, vertigo and limbs numbness. Suffering from these recurrent symptoms, she underwent many examinations but all the survey could not tell an answer. Finally, by the presence of porphyrin in urine testing, the diagnosis of porphyria was established. Other associated laboratory data showed elevated 24-hr prophobilinogen and Delta-aminolevulinic acid (Delta-ALA) levels in urine. Without a genetic study, the typing was recognized as acute intermittent porphyria based on the clinical presentations. In addition to porphyria, the patient also had past history of hyperthyroidism and depressive disorder with pseudo-hallucination. She never received any endovascular intervention or implantable vascular access devices.

The IVC thrombosis event was in the same year of diagnosis. It was before the diagnosis of porphyria, but after the associated symptoms onset. The patient visited emergency department with complaint of chest pain, accompanied with cold sweating, dizziness, orthopnea and paroxysmal nocturnal dyspnea in July, 2017. Although cardiac enzyme elevation was absent, with those symptoms suspicious for acute coronary syndrome, she received Thallium-201 myocardial perfusion imaging, which showed probably myocardial ischemia at the anterior segment of left ventricle. The following cardiac catheterization revealed coronary vasospasm. During the vascular access, inadvertent punctures of right femoral vein were present in the multiple puncture attempts to right femoral artery. After the procedure, the hemostasis was achieved by immobilization and sandbag compression for 6 hours.

During afterward admission days, soreness over lateral part of right buttock and upper thigh developed, also, ecchymosis at suprapubic area. In the computed tomography (CT) scan of pelvis, procedure changes and small hematoma in the right external iliac region of the pelvic cavity were observed. Moreover, the CT scan disclosed thrombosis in the right common iliac vein and the proximal IVC, more specifically, sub-renal IVC (Figure 1). The blood clot was monstrous, largely extending. The next day, IVC filter was implanted for prevention of circulating embolism. The data of coagulation profile, including protein C, protein S, prothrombin time and activated partial thromboplastin time, was all in normal ranges. The patient took Rivaroxaban for 4 months as maintenance anticoagulation therapy of venous thrombosis.

In the CT scan 6 weeks later to the IVC filter implantation, the IVC and right common iliac vein thrombosis disappeared (Figure 2A). The patient returned to outpa-

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tient clinic regularly on time, without any thrombosis or embolic events reported. The intermittent abdominal pain during this period of time was contributed to porphyria. The IVC filter was successfully removed 4 months later, in November, 2017. The removal was through right internal jugular vein antegradely under echo guidance.

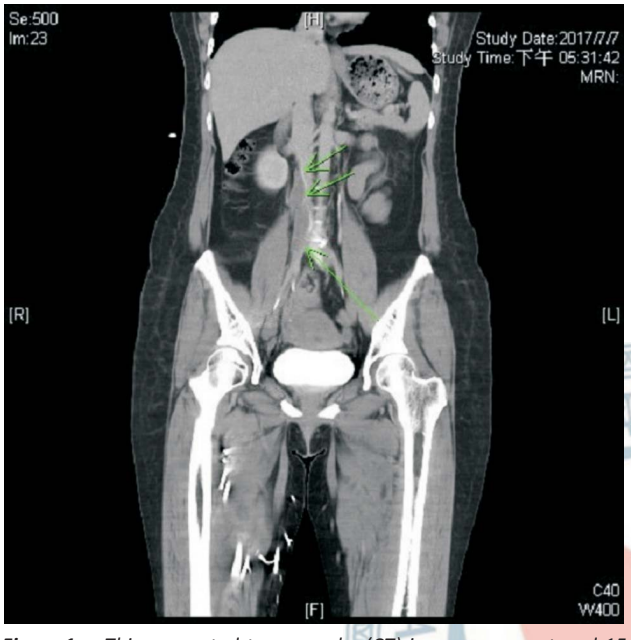

Figure 1. *This computed tomography (CT) image was captured 15 days after the cardiac catheterization. It presents large inferior vena cava (IVC) thrombosis (green arrows) extending from sub-renal level to right common iliac vein.*

No any IVC injury was noted during the procedure. The CT 2 weeks after the IVC filter removal indicated that the IVC and right common iliac vein remained patent (Figure 2B).

Intermittent acute attacks of porphyria occurred in the following years. Hemin therapy was even applied to her. She had no record of invasive procedure or a thrombosis event afterward.

DISCUSSION

Porphyria is a group of rare, inherited diseases with enzyme defects in the heme biosynthesis pathway.[1-5] Partial absence or reduced activity of specific enzyme causes accumulation of intermediate prior to the synthetic step of that enzyme, and the following clinical symptoms.[2-4]

The classification of porphyria bases on involved organs and clinical manifestations, also the affected gene and biochemistry features.[2,3,5] The majority of heme production is in liver and bone marrow, thereby porphyria is either hepatic or erythropoietic designation.[2,5,6] By genetic traits and clinical courses, they could be further classified.[2,6] The pathophysiologic mechanisms of these toxic heme synthesis precursors in types of porphyria resulting in various clinical symptoms are not fully

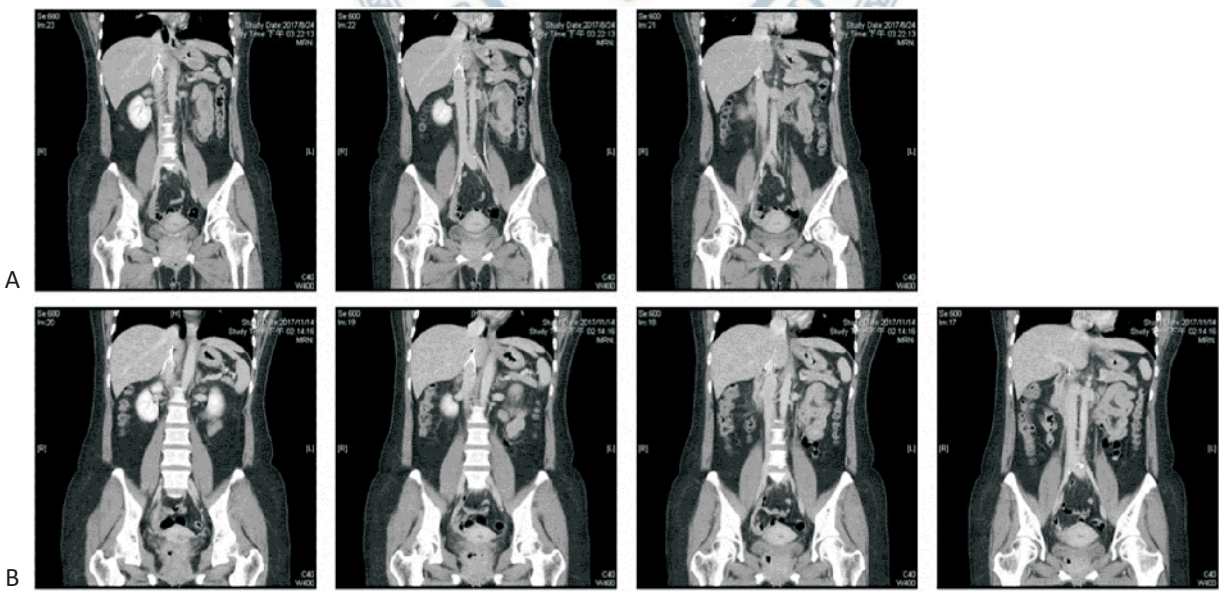

Figure 2. *(A) No blood clot is observed in the computed tomography (CT) image 6 weeks later to the inferior vena cava (IVC) filter implantation. (B) The follow-up CT image 2 weeks after the IVC filter removal presents patent both IVC and right common iliac vein.*

understood.[2] Diagnosis of porphyria depends on biochemical and genetic testing.[2-6]

The acute attacks could be triggered by certain factors, lasting for days to weeks.[1,3,4] By affecting automatic, central and peripheral nervous systems, accumulated heme precursors are responsible for multiple neurological symptoms.[1] Neurologic origin abdominal pain without response to narcotic agents is the most common symptom, possibly accompanied with nausea, vomiting and constipation.[1,3,4,6] Other autonomic symptoms, like hypertension, tachycardia or episodic sweating, may be observed.[1,3,4,6] Including muscle weakness, paresis and sensory neuropathy, peripheral neuropathy is also common.[1,6] As for porphyria-induced encephalopathy, the clinical manifestations could be seizure, syndrome of inappropriate antidiuretic hormone, and even posterior reversible encephalopathy syndrome.[1,3,4] Neuropsychiatric symptoms sometimes take part in, such as depression, anxiety, insomnia, hallucination and delusion.[1,3,6] Some types of porphyria are due to overproduction of photosensitizing precursors and cause photocutaneous manifestations.[2-4]

Thrombosis is not an usual presentation in patients with porphyria. However, an IVC thrombosis event in a case with porphyria after portacath implantation was reported in 2005.[7] Similar to our case, it was a young-age female patient with history of acute intermittent porphyria receiving invasive intravascular procedure. IVC thrombosis developed around the portacath, below the renal vessels level and extending into the iliac vein.

In a retrospective study, increased liver artery thrombosis complication incidence of liver transplantation in patients with porphyria was reported, which was up to 40%.[8] There were 10 patients receiving liver transplantation due to recurrent acute attack and severe symptoms caused by porphyria. As the complication rate was about 5% in the population undergoing liver transplantation, in the study, liver artery thrombosis occurred in 4 out of 10 patients diagnosed with porphyria.[9,10] Besides, it was mentioned that central venous catheter indwelling in porphyria patients frequently leaded to vascular thrombosis.

Inadvertent punctures of adjacent vein, immobilization and prolonged sandbag compression for hemostasis are not hardly seen in daily practice, which might provoke venous thrombosis development in our case. However, IVC thrombosis is pretty rare in absence of congenital or acquired IVC abnormalities. As a porphyria case, a rare disease, the patient was diagnosed with IVC thrombosis in high level of severity, which is another disease with very low incidence. Coincidentally, a case with similar clinical course was ever reported. We do not describe the coincidence of these two diseases as occasional. The correlation of porphyria and coagulation is not clear so far and only sporadic reports involved. However, IVC thrombosis occlusion is dangerous and early intervention is crucial. We consider thrombotic events should be minded in caring of porphyria cases in the future, especially those cases receiving invasive intravascular procedures.

LEARNING POINT

This case report highlights the awareness of thrombotic events in porphyria patients. Porphyria patients might have increased venous thromboembolism risks. Thrombotic complications should be paid attention to in porphyria patients receiving endovascular intervention. Radiologic imaging has become highly relevant to detection of possible thrombotic complications and planning of intervention.

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DECLARATION OF CONFLICT OF INTEREST

All the authors declare no conflict of interest.

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