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ΑΡΧΕΙΑ ΕΛΛΗΝΙΚΗΣ ΙΑΤΡΙΚΗΣ 2019, 36(1):143

Internal Medicine Quiz – Case 22

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A 52-year-old male Greek, non-smoker, with a medical history of non-alcoholic fatty liver disease was admitted to our hospital because of a diffuse abdominal pain, progressively worsening for 10 days, accompanied by nausea and some diarrhea. Physical examination revealed a painful abdomen with decreased intestinal motility and a palpable spleen. Blood examinations revealed normal values of hemoglobin (15g/dL), hematocrit (45%) and platelets (235.000/μL), polymorphonuclear leukocytosis (WBC [white blood cells]: 12.100/μL), elevated levels of serum transaminases (AST: 42 IU/L and ALT: 76 IU/L), C-reactive protein [CRP] (8.6 mg/L; normal range: 0–5 mg/L) and homocysteine (18.5 μmol/L; normal value <15 μmol/L), and low levels of folate (2.1 ng/mL; normal range: 3–20 ng/mL). An abdominal ultrasound revealed a moderate splenomegaly (18.5 cm) and a trace of fluid in the Morrison pouch. An abdomen/pelvis computed tomography (CT) scan showed portal vein thrombosis (PVT) extending into superior mesenteric and splenic veins (fig. 1). The liver parenchyma was normal in appearance. Investigation into possible causes of non-cirrhotic, non-malignant, acute PVT revealed three mutations: (a) Factor V Leiden (heterozygous G1691A), (b) *MTHFR* gene (homozygous C677T, heterozygous A1298C) and (c) Janus kinase-2 (JAK2V617F).

Comment

Acute PVT is an unusual thrombotic condition associated with significant morbidity and mortality. The local causes of PVT include liver cirrhosis, abdominal infections and inflammatory conditions, inflammatory bowel disease, abdominal malignancies and portal vein injury. Among inherited thrombophilic disorders, prothrombin G20210A mutation appears to be prominent in the pathogenesis of PVT. Among acquired thrombophilic disorders, the most common causes of PVT are associated with JAK2 V617F mutation myeloproliferative disorders (MPD), such as polycythemia vera, essential thrombocythemia and idiopathic myelofibrosis. Because acute PVT might even be the first manifestation of a MPD (overt or latent form), the identification of JAK2 mutation (major diagnostic criterion for MPD diagnosis) is necessary. Anticoagulation therapy is the cornerstone of PVT treatment. Our patient initially received low-molecular-weight heparin which was replaced after three weeks by acenocoumarol targeting the international normalized ratio (INR) at a range of 2.5–3. Due to the presence of prothrombotic disorders in our patient, we decided on lifelong intake of acenocoumarol. Changes in lifestyle were also suggested, such as lifelong intake of L-methylfolate supplement to control the methylenetetrahydrofolate reductase (MTHFR) mutation gene-induced hyperhomocysteinemia. Imaging follow-up with Doppler ultrasonography and abdomen/pelvis CT scan after 3 and 6 months, respectively showed recanalization of the splenoportal axis. Upper gastrointestinal endoscopy was normal. Regarding the JAK2 gene mutation, the patient was referred to the hematology clinic of a tertiary hospital. Two years later, polycythemia vera was diagnosed in our patient.

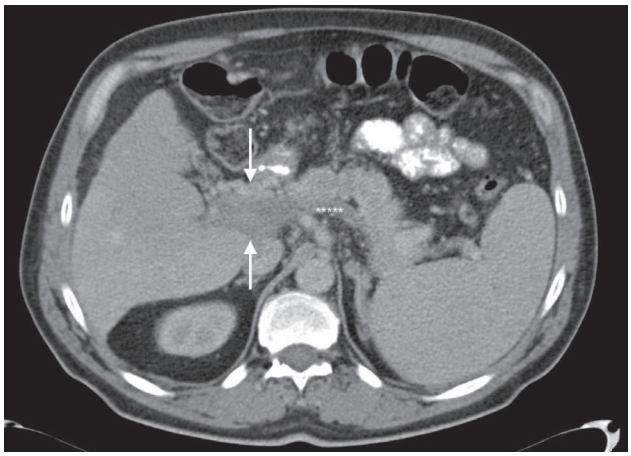

Figure 1. Abdomen/pelvis computed tomography (CT) scan. Between white arrows a dilated, thrombosed portal vein is shown with multiple vascular collaterals in keeping with portal vein cavernous transformation. The white asterisks indicate the splenic vein which is also thrombosed. The superior mesenteric vein thrombosis is not shown in this figure.

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Diagnosis: *Non-cirrhotic, non-malignant, acute PVT due to polycythemia vera*