Venous thrombosis-related abdominal pain

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To the Editor;

An unexpected abdominal thrombotic event in a patient is cause for following evaluation of an underlying hypercoagulable state. Portal venous thrombosis (PVT) and Budd-Chiari syndrome (BCS) frequently result from multiple different factors including cirrhosis, intraabdominal sepsis, procoagulant states, and myeloproliferative neoplasms (MPN) (1). MPN are responsible for 50% cases of BCS and 35% cases of PVT in western series (2). The JAK2 V617F mutation is a point mutation in the Janus kinase 2 (JAK2) tyrosine kinase that is variably present in chronic MPNs. We report the case of a 55-year-old woman with spontaneous, unprovoked abdominal PVT; and thus we want to demonstrate how testing for the JAK2 V617F mutation was helpful in unmasking an induced coagulability.

She presented to our hospital with one month of upper abdominal indefinite pain and history of unexamined enlarged spleen. She had no known trauma, nausea-vomiting, constipation or diarrhea. She denied a history of medically confirmed liver or blood diseases; and fatigue, fevers, night sweats, weight loss and chills. The initial Doppler ultrasonographic examination revealed subacute venous thrombotic obstruction in portal vein, and the upper gastrointestinal endoscopy showed doubtful esophageal varices. A computed tomography scan of the abdomen-pelvis was performed, demonstrating a heterogeneous appearance of the liver, massive splenomegaly, no ascites and no mass or lymphadenopathy. Her medical history was not notable for disease or medication. She had never smoked and drank. Laboratory findings revealed a white blood cell count of 7.3 x 10⁹/L, hemoglobin of 13.5 gr/dL, platelet (PLT) count of 438 x 10⁹/L. Alanine aminotransferase, aspartate aminotransferase, gamma glutamyl transferase, alkaline phosphatase, total and direct bilirubin, total protein and albumin, kidney function tests and electrolytes, and erythropoietin levels were normal. The patient was evaluated for a hypercoagulable state, including tests for prothrombin 20210 and factor V Leiden mutation, lupus anticoagulant and antiphospholipid antibodies were negative. Levels of anti-thrombin, protein C and S were normal. But the result of JAK2 V617F mutation was heterozygous positive. Mild leukoerythroblastosis was found in peripheral blood smear test. Bone marrow aspiration and biopsy examination showed proliferation mainly of the megakaryocytic lineage with increased numbers of enlarged, mature-immature and atypical megakaryocytes with marked reticulin fibrosis. Complete blood counts obtained one year ago were notable for thrombocytosis (PLT count was 480 and 500 x 10⁹/L for two times). On the basis of these findings, the patient’s condition was diagnosed as an underlying chronic MPN-associated myelofibrosis (idiopathic myelofibrosis). The medication including low molecular weight heparin and acetyl salicylic acid with hydroxyurea was started. Allogeneic stem cell transplantation as a treatment choice was planned to discuss.

The chronic MPNs are a group of clonal neoplastic malignancies that result in the overproduction of one or more normally differentiated myeloid lineages (3). JAK2 V617F mutation has been implicated in 90% of cases of polycythemia vera, and up to 30% of cases of essential thrombocytemia and idiopathic myelofibrosis. Natural history of the MPNs is...
marked by thrombo-hemorrhagic complications and a tendency to transform into myelofibrosis and acute leukemia (4). MPN now constitutes the most common cause of abdominal venous thrombosis (5). The clinical features of primary myelofibrosis are variable and include anemia, leukopenia or leukocytosis, thrombocytopenia or thrombocytosis and extramedullary hematopoiesis, most commonly causing hepatomegaly and symptomatic splenomegaly (6). In the present case, routine hemogram and biochemical parameters were in the normal range; splenomegaly and peripheral blood smear test results were consistent with the disease only. Based on this, we would like to draw attention to the importance of physical examination and peripheral blood smear.

Finally, this case shows the importance of investigation of JAK2 V617F in patients presenting with abdominal venous thrombosis, even in the absence of apparent hematologic abnormalities.

REFERENCES