SPLACHNIC VEIN THROMBOSIS AND MYELOPROLIFERATIVE SYNDROMES. THE ROLE OF JAK2V617F MUTATION

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Abstract text

1.Background:Myeloproliferative diseases(MPDs) are shown to have an increased risk of thrombotic complications such as splachnic vein thrombosis(SVT).Mutations on JAK2 pathway are thought to play key role on such thrombotic complications. 2.Aims:The focus of the current work is to evaluate the risk of SVT in MPDs patients and its colleration with the mutation JAK2V617F. 3.Methods:Patients with non-cirrhotic, non-cancer related SVT and with clinical or laborating findings suggesting MPD were assessed for the presence of JAK2V617F mutation. We suspected that normal or light increased platelet count might mask MPDs (portal hypertension-hypersplenism, occult bleeding).Assessment for hematological pro-coagulant conditions included factor V Leiden, antithrombin III, protein C, protein S, homocysteine, MTHFR mutation, prothrombin gene mutation PT20210A, anticardiolipin antibodies and lupus anticoagulant. Paroxysmal nocturnal hemoglobinuria was screened using standard flow cytometry techniques. Patients with known history of pylephlebitis were excluded. SVT was confirmed with computerized tomography and abdominal doppler ultrasound. SVT was characterized as chronic if there was evidence of intra-abdominal venous collaterals, carvenous transformation of the portal vein, or signs of portal hypertension. 4.Results:In the study 14 patients were included. The median age at the time of diagnosis was 50.71 years (range, 21-78) and 57% were male. All patients had chronic SVT, 64% had PVT and the rest were diagnosed with BCs. Every patient underwent bone marrow biopsy: polycythemia vera(PV) 4 patients, essential thrombocytosis(ET) 7 patients, primary myelofibrosis(PMF) 3 patients. JAK2V617F was analyzed in 12/14 patients and was positive in 100%. Inherited thrombophilia was not found. Acquired thrombophilia was mentioned in two patients. A woman with Budd-Chiari syndrome(BCs) who was provided oral contraceptive pills, and a man with portal vein thrombosis(PVT) post-splenectomy. Patients with BCs had mean age 43.2 years(range, 35-56) and 60% were female. Three were diagnosed with PV, 1 ET and 1 PMF. One patient died after 17 years and one was scheluded for liver transplantation after 6 years. The other three patients had no signs of ascites or portal hypertension in a six-year follow up. Patients with PVT had mean age 54.8 years(range, 21-78) and 67% were male. Six were diagnosed with ET, 2 PMF and 1 PV. On admission 5 patients had esophageal/gastric varices whereas 89% patients had splenomegaly. Five patients had also evidence of superior mesenteric vein thrombosis. Nobody died. All of the patients have signs of portal hypertension. Mean time of follow up is 1.8 years(range, 0.2-6). All patients were managed with routine anticoagulation therapy from diagnosis. Three patients had indications for decompressive procedures such as TIPS, all in the group of BCs. 5.Summary/Conclusions:SVT is frequent presenting complication of undiagnosed MPDs.In patients with SVT, portal hypertension is a virtually constant feature. The resulting hypersplenism and hemodilution decrease the accuracy of blood cell counts for MPD diagnosis. The atypical peripheral blood picture in the setting of SVT has led to a variety of denominations such as "latent” MPDs. In our study, all patients with MPD and SVT were positive for the mutation JAK2V617F.The presence of this mutation may predict a more aggressive phenotype with an increased risk of thrombosis.