Case Report

Hyperhomocysteinemia with Simultaneous Superior Mesenteric, Portal and Splenic Vein Thrombosis in a Young Adult

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Abstract:
Hyperhomocysteinemia is a known risk factor for atherosclerosis leading to venous and arterial thrombosis at an early age. The present report presents an unusual case of multiple and simultaneous venous thrombosis in a patient with hyperhomocysteinemia. A 36-year-old man presented with acute pain in the right side of abdomen. Computed tomography of the abdomen indicated thrombosis in right branch of portal vein, intra and extra hepatic portal vein, superior mesenteric vein and splenic vein. After evaluation it was found that patient had hyperhomocysteinemia.

Keywords: venous thrombosis, atherosclerosis, Computed tomography, hyperhomocysteinemia

Introduction:
Hyperhomocysteinemia is a known risk factor for atherosclerosis leading to venous and arterial thrombosis at an early age. Different organ systems can get involved and manifest as stroke or ischemic heart disease or peripheral vascular disease (1). A 36-year-old man presented with acute pain in the right side of abdomen lasting three days. He was a known alcoholic and non-smoker. He had no history of diabetes mellitus, hypertension, hyperlipidaemia or prior episode of vascular thrombosis. A physical examination at the time of admission revealed normal findings. There were no signs of peritoneal irritation.

Laboratory tests revealed normal findings [haemoglobin (Hb): 15.1 g/L; mean corpuscular volume (MCV): 98fL; and mean corpuscular haemoglobin: 33.3pg]. Total bilirubin (0.71 mg/dL; reference range: <1.2 mg/dL), amylase (33 U/L; reference range: 0–86 U/L), aspartate aminotransferase (83 IU/L; reference range: <40 IU/L), alanine aminotransferase (17.7 IU/L; reference range: <42 IU/L), alkaline phosphatase (265 IU/L; reference range: <270 IU/L) and γ-glutamyltransferase (28 IU/L; reference range: 7–45 IU/L). Activated partial thromboplastin time (TTPa): 34.8 seconds (reference range: 25–34 seconds), prothrombin time: 15.5 seconds (reference range: 10–15 seconds), international normalized ratio (INR): 1.3, Creatinine (1.13 mg/dL; reference range: 0.6–1.2 mg/dL).

The result of an abdominal ultrasonography and upper GI endoscopy was normal. An intravenous contrast-enhanced computed tomography of the abdomen, performed because of persistent pain, indicated thrombosis in right branch of portal vein, intra and extra hepatic portal vein, superior mesenteric vein and splenic vein (figure 1-3).

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Image 1 and 2: Contrast Enhanced CT abdomen showing filling defect in superior mesenteric vein suggestive of
thrombus (Depicted by arrow)

The patient was treated with Inj. Enoxaparin (0.6 mg subcutaneously every 12 hours) overlapped with oral anticoagulant tab. Warfarin which was gradually titrated to achieve an INR between 2 and 3. Since multiple simultaneous thrombosis were found investigation of thrombophilia was done. Anti-thrombin III (Antigenic): 17 (reference range: 17-30), factor V leiden mutation detection by PCR was negative, protein C: 86.9 (reference range: 70-150), protein S: 75.5 (reference range: 70-150, suggesting normal. His fasting serum homocysteine level was high (49.5 µmol/L; reference range: <15 µmol/L).

A diagnosis of hyperhomocysteinemia as a cause of venous thrombosis was made and patient was started on pyridoxine, vitamin B 12 and folic acid supplements. Pain in abdomen subsided after treatment and was discharged on day 7 and is awaiting follow up.

Discussion:

Homocysteine is derived from essential amino acid methionine. It is primarily metabolized either via re-methylation to methionine, or via irreversible transulfuration to cysteine. The enzyme methionine synthase remethylates homocysteine to methionine. Vitamin B12 (cobalamin) and dietary folate are essential cofactors for methionine synthase. Interruption of metabolism can result in Hyperhomocysteinemia. It is a rare disorder with an incidence of 1 in 2, 00,000 to 10, 00,000 population.(5)

The association of homocysteine excess and arteriosclerotic vascular disease was established in 1969, by McCully.(6) The pathogenesis of arteriosclerotic occlusive disease due to Hyperhomocysteinemia is unknown. Oxidative injury to the endothelium and underlying vascular matrix, and proliferation of the vascular smooth muscle has been proven in some studies as the cause for occlusive disease.(7)

Pyridoxine, vitamin B 12 and folic acid supplements are given in patients of Hyperhomocysteinemia. These three in combination reduces the homocysteine levels and also provide some clinical benefits.

Conclusion:

While evaluating cases of arterial and venous thrombosis, especially in young patients, hyperhomocysteinemia should be a prime consideration. It is one of the most common etiologies among the long, exhaustive thrombophilia panel. Furthermore ordering a thrombophilia panel in rural and low social-economic strata does not seem practically feasible. We would advise serum homocysteine level to be considered as a primary test before subjecting the thrombophilia profile in suspected cases of venous thrombosis wherever appropriate. Needless to emphasize other common conditions should be ruled out.

References: