Regenerative nodular hyperplasia, portal vein thrombosis and primary myelofibrosis: an unusual triple association

Key words: Regenerative nodular hyperplasia. Portal vein thrombosis. Primary myelofibrosis. JAK2 gene.

Dear Editor,

Portal vein thrombosis (PVT) or regenerative nodular hyperplasia (RNH) have been reported in cases of primary myelofibrosis (PM). The co-occurrence of all three conditions has never been previously reported to the best of our knowledge.

Case report

The condition initially developed when the patient was 27 years of age and presented as a fundal variceal bleeding. A portal vein cavernoma was identified and the results of the hypercoagulability study were normal. A liver biopsy identified evidence of RNH. At age 36, the patient was readmitted due to vomiting, abdominal pain and a mild fever. A computed tomography (CT) scan identified intestinal wall thickening and a new-onset occlusive thrombosis in the superior mesenteric vein (Fig. 1). The patient tested positive for the V617F mutation in the JAK2 gene. A bone marrow study was consistent with PM, and hypocoagulation with enoxaparin was subsequently initiated concurrently with sequential hydroxyurea-ruxolitinib therapy. The patient had repeated sub-occlusive attacks related to a partial stenosis of the middle ileum. Three months later the patient required an emergent segmentary ileal resection due to acute complications from an inviable intestine.

Discussion

RNH develops as an adaptive and pathological response to obliterator venopathy (2), and the underlying thrombophilia may act as the required trigger (3). Intrahepatic myeloid metaplasia and extramedullary hematopoiesis are both reversible conditions with a specific treatment and have also been associated with PM (4). PVT risk is higher in patients with RNH compared to patients with cirrhosis; the relationship between PM and thrombosis is also well known (2). Taking into account the high incidence of PVT in RNH and the high rate of thrombophilic disorders in both conditions, it is possible that they represent different presentations of a single disorder. The take-home message from this case report is that, in addition to routine hypercoagulability tests, myeloproliferative disorders should be promptly and appropriately screened in PVT cases (5).

Fig. 1. IV contrast-enhanced abdominal CT scan. The arrow shows a filling defect in the superior mesenteric vein, which corresponds to occlusive thrombosis associated with a right colonic hypoperfusion.
References


