Case Report: *JAK2* Mutation in a Patient With Splanchnic Venous Thrombosis

Nour A. Haidar[1] , Youssef Jounblat [2], Karine Salignon [2], Helene Paradis [2], Willy Vaillant [2]

[1]Saint Louis Hospital, Paris, France. [2]Hopital AUCH, AUCH, France .

Centre Hospitalier d'Auch EN GASCOGNE — Soigner & prendre Soin

Splanchnic venous thrombosis (SVT) is known to be the most frequent underlying prothrombotic factor. frequently associated with prothrombotic conditions such as myeloproliferative neoplasms (MPNs), particularly JAK2-V617F mutation-positive polycythemia Vera (PV) and Essential thrombocythemia (ET) (1, 2).

Case presentation

- We present a case of a 63-year-old man who presented to the ER in 2013 with a liver function test abnormality (ASAT 3.5 N, ALAT 4N, GGT 2.5 N, ALP N) and with abdominal pain in the right upper quadrant.
- A CT scan showed a superior mesenteric vein thrombosis (SVT) affecting branches of the portal trunk, probably of idiopathic origin. A gastroscopy appeared normal, while an ultrasound endoscopy detected the SVT and a floating clot. An abdominal MRI showed hepatomegaly of 3.5 cm below the costal margin with dilatation of the portal vein, no metastases or lesions.
- Initiation of LMWH .
- A bone marrow biopsy was performed, which revealed no proliferative bone marrow changes.
- Further genetic testing revealed a positive *JAK2-V617F* mutation. Thrombophilia screening results were negative for ANCA; anti-phospholipid, anti-ENA, anti-cardiolipin, and anti-beta 2 glycoprotein 1 antibodies; factor V Leiden; protein C; protein S; and prothrombin factor II mutation.
- Control of blood tests showed continued elevated platelets and WBC counts.
- In January 2023 Investigations revealed a blood test showed Hb 17.2 g/dL, HCT 51.5%, platelets 567,000, AST 33 UI/L , ALT 37 UI/L, alkaline phosphatase 53 UI/L and total bilirubin 19.7 mg/l with a CRP and ferritin in the normal range.
- The patient was treated with aspirin and hydroxyurea 1 tablet/day. The patient remained stroke free and platelet levels were normal throughout the 1-year follow-up period.

DISCUSSION

- This case report describes the discovery of SVT involving branches of the portal trunk. Diagnosis often requires a high index of suspicion due to nonspecific symptoms and overlapping laboratory findings, necessitating imaging modalities such as CT and MRI (1, 2).
- Identification of the JAK2-V617F mutation in this case was pivotal as it confirmed the underlying MPN. JAK2 mutations are prevalent in SVT patients with MPNs and play a crucial role in risk assessment and disease management due to their association with thrombotic events (1, 3).
- WHO 2016 diagnostic criteria for ET based on persistent thrombocytosis and bone marrow findings. Treatment included aspirin for thromboprophylaxis and hydroxyurea to control platelet counts, leading to a favorable clinical outcome without thrombotic complications during follow-up (4).

CONCLUSION

- In our case of a 63 year-old male with JAK2 V617F mutation , where this mutation is frequently found in patients with SVT even when many do not have an overt myeloproliferative disorder (MPD).
- Splanchnic venous thrombosis (SVT) encompasses thrombosis of the hepatic, portal, and mesenteric venous systems. It can either be an initial indication or a later complication of myeloproliferative neoplasms (MPN). Predicting which patients will develop overt MPN at the time of presentation is challenging. Nevertheless, both overt and latent MPNs significantly increase the risk of thrombosis in the portal, mesenteric, and hepatic regions (5) .
- Several studies suggest that anticoagulation therapy may lower bleeding risk by : Promoting vein recanalization or Decrease intravascular pressure in the splanchnic circulation .
- Reducing pressure in gastrointestinal varices (6-7)
- Myeloproliferative disorders (MPDs), whether overt or latent, represent a main intrinsic factor for the development of thrombosis in the portal, mesenteric, and hepatic areas (8-9-10-11) .
- Long-term management of SVT in MPNs requires a multidisciplinary approach focusing on anticoagulation, risk stratification based on genetic and clinical factors, and regular monitoring for disease progression and thrombotic events (12).

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