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Essential Thrombocythemia and Ulcerative Colitis: Unusual Association

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Abstract

Essential thrombocythemia is classified as a chronic myeloproliferative disorder characterized by the overproduction of platelets stemming from a megakaryocytic clone. The diagnosis primarily relies on bone marrow biopsy findings and the detection of the JAK2 V617F mutation, after the exclusion of secondary thrombocytosis due to conditions such as inflammation, hemolysis, infection, and iron deficiency. On the other hand, Ulcerative colitis represents an inflammatory disorder of the colon. The diagnosis of ulcerative colitis is established through clinical assessment, endoscopic examination, and histological criteria, without a discernible alternative etiology. The concomitant occurrence of these two conditions is infrequent. We present the case of an 85-year-old patient with a history of essential thrombocythemia who exhibited gastrointestinal symptoms characterized by alternating episodes of diarrhea and constipation. A subsequent colonoscopy accompanied by a biopsy revealed histological features consistent with ulcerative colitis. The patient was administered cytoreductive therapy in combination with mesalazine, resulting in favorable outcomes. Current literature addressing this association is limited, indicating the need for further investigative studies to elucidate the causal relationships between these two pathologies and to achieve improved therapeutic management strategies.

Keywords

Thrombocytosis, Essential Thrombocythemia, Rectal Bleeding, Ulcerative Colitis, Case Report

1. Introduction

Essential thrombocythemia is an acquired myeloproliferative disorder characterized

by a persistent elevation in platelet count, with a predisposition to both thrombosis and hemorrhage [1]. Ulcerative colitis is a chronic inflammatory condition affecting the colon [2].

Studies show that patients with IBD are at increased risk of various chronic conditions, such as cardiovascular diseases, osteoporotic fractures, and other immune-mediated diseases. In addition, the risk of gastrointestinal cancers is increased, as is the risk of some extra-intestinal malignancies, including hematological cancers [3] [4].

An association between hematological cancers and inflammatory bowel disease (IBD) has previously been suggested, but the risk of IBD in patients with myeloproliferative neoplasms (MPNs) is unknown [5].

We present a case of a patient diagnosed with essential thrombocythemia who developed ulcerative colitis during her clinical follow-up.

2. Case Presentation

This is an 85-year-old patient followed for 30 years for depression on antidepressant treatment based on Amilsupride 50 mg/day + Mianserin 30 mg/day, operated 15 years ago for an undocumented digestive problem. The patient has no other significant medical history, including no diabetes, no hypertension, and no cardiovascular disease.

The patient presented one year before her admission a deterioration of the general state with an unencumbered weight loss justifying her consultation in the internal medicine department in 2015. The general examination found a conscious patient with GCS 15/15th stable on the hemodynamic and respiratory plan, normotensive at 130/69 mmHg, normocardium at 80 beats/min, à normal respiratory rate at 16cycles/min and afebrile at 37.6, slightly discolored conjunctiva, urine output preserved. Clinical examination found a midline laparotomy scar consistent with old surgery. Furthermore, the remainder of the examination showed no palpable splenomegaly or lymphadenopathy. The blood count (CBC) showed thrombocytosis at 1,038,000 elements/mm^3 with smear anisochromia without blasts. The remainder of the count showed normochromic, a regenerative normocytic anemia with Hemoglobin at 11.4 g/dl VGM 93 u3, reticulocytes at 57,000, neutrophils at 4874 elements/mm^3 and lymphocytes at 2394 elements/mm^3 (**Table 1**).The other secondary causes of thrombocytosis were eliminated: ferritin at 35, no context of inflammation; CRP at 10 and VS at 25 mm, no hemolysis with total bilirubin at 5 mg/l and indirect bilirubin at 3 mg/l haptoglobin at 0.68 g/l and LDH at 327 IU/L. No infection with serology hepatitis B, C, and HIV IDR negative to tuberculin negative. The abdominal ultrasound didn't show any splenomegaly (**Image 1**). The CT scan of the Cervico-Thoraco-Abdomino-Pelvic region was performed objectifying a thickening of the recto-sigmoid junction of 11 mm (**Image 2**). The Colonoscopy with polyp biopsy was performed showing a well-differentiated ulcerated and inflammatory polyadenoma (**Image 3**).

The diagnosis of essential thrombocythemia (ET) was established based on the

Table 1. The results of the patient's biological assessment.

	Patient results	References values
Hemoglobin	11.4 g/dl—7.12 mmol/l	12 - 16 g/dL—7.5 - 9.9 mmol/L
MCV (Mean Corpuscular Volume)	93 um^3—93 Fl	80 - 98fL—80 - 98 μm^3
Reticulocyte rate	57,000/ul—57 G/L	80 - 98 fL—80 - 98 μm^3
neutrophils	4874 element/mm^3—4000/ul	1500 - 7000/µL—1500 - 7000/mm^3
Platelets	1,038,000 element/mm^3—103 G/L	150,000 - 400,000/µL—150 - 400 G/L
Ferritin	35 mg/l	30 - 280 mg/l
CRP (C-reactive protein)	10 mg/l—95 nmol/L	0 - 5 mg/L—0 - 47 nmol/L
Erythrocyte Sedimentation rate at first hour	25 mm	F < 20 mm M < 20 mm
Total bilirubin	5 mg/l—8.3 mmol/l	12 mg/l—20 mmol/l
Indirect bilirubin	3 mg/l—5.4 mmol/l	10 mg/l—18 mmol/l
Haptoglobin	0.68 g/l	0. 3 - 2 g/l
LDH	327 UI/l	140 - 245 UI/l

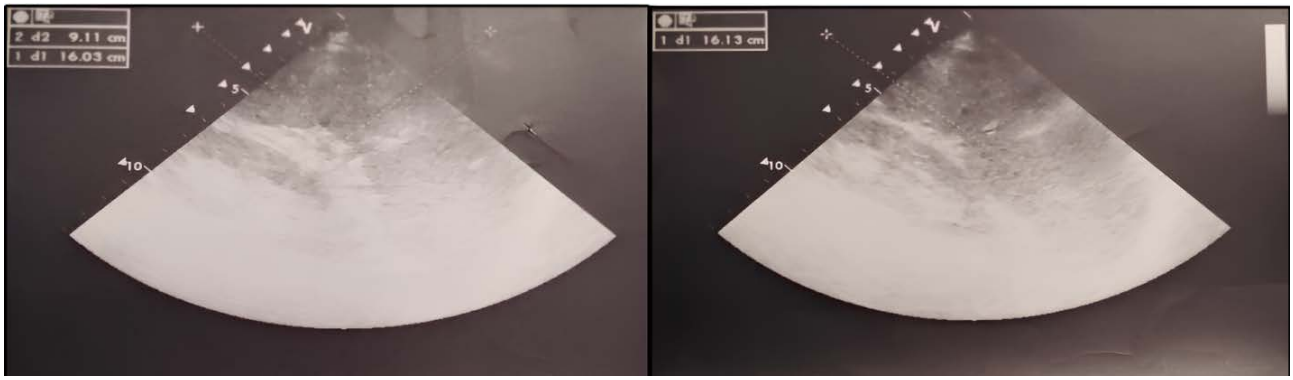

Image 1. Normal abdominal ultrasound.

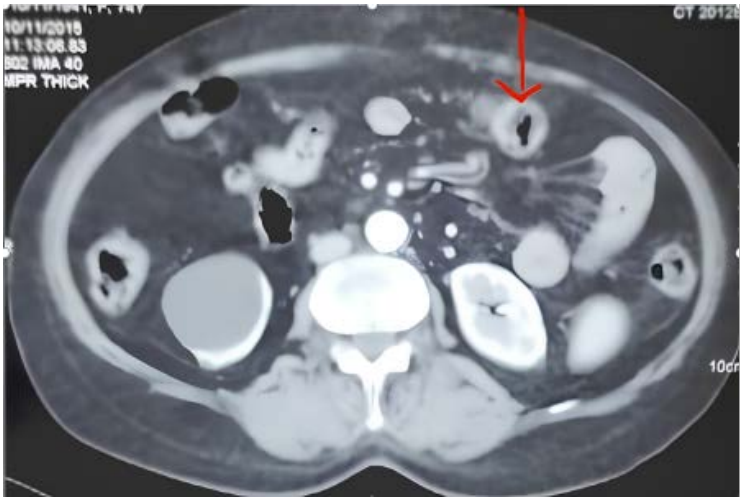

Image 2. The CT scan of the Cervico-Thoraco-Abdomino-Pelvic region objected to a thickening of the recto-sigmoid junction of 11 mm.

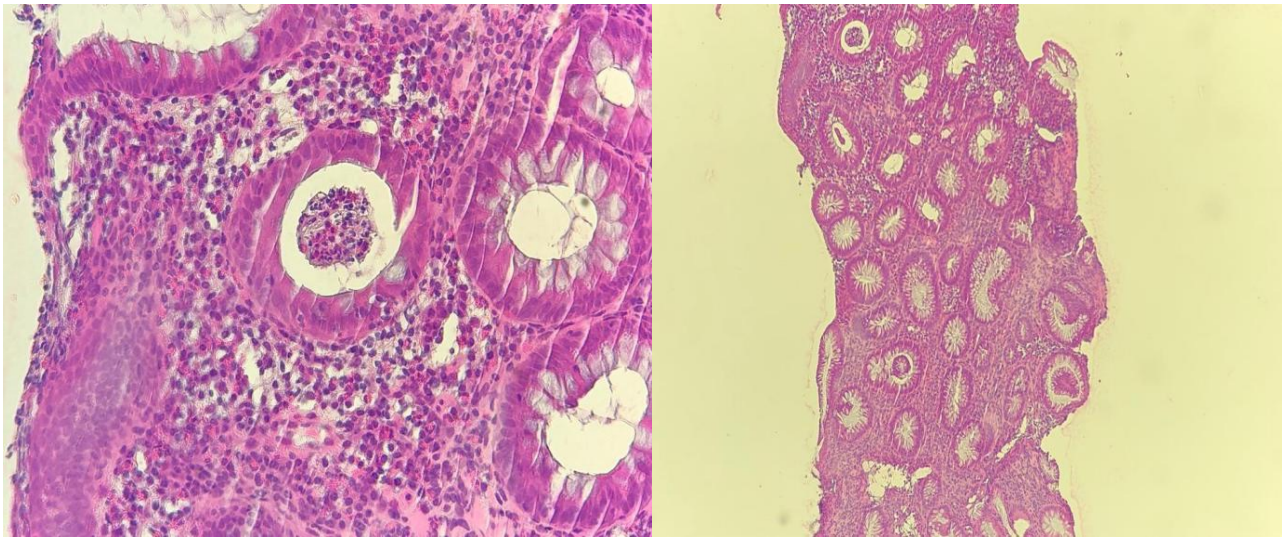

Image 3. Histological aspect of a rectal biopsy showing ulcerative colitis.

presence of the WHO major criteria, including thrombocytosis, the positive JAK2 V617F mutation, the bone marrow findings (a moderately rich polymorphic marrow with no signs of specific inflammation or malignancy), and the exclusion of other causes of thrombocytosis. The patient was treated with cytoreducer treatment (hydroxyurea) with good progress during her follow-up.

In 2016, the patient consulted again for rectal bleeding with alternating diarrhea and constipation. The biological assessment showed a thrombocytosis at 825,000 elements/mm^3 with albumin at 37 g/l, and a copro parasitological examination of the altered stools returned normal. A second colonoscopy combined with a biopsy showed a hyper vascularized fibro-oedematous chorion infiltrated by inflammatory lymphoplasmacytic elements, polynuclear neutrophils, and eosinophils suggesting ulcerative colitis.

The diagnosis of ulcerative colitis was established based on a combination of the clinical presentation including the diarrhea and the rectal bleeding, the endoscopic aspect, and histological findings.

This time the patient was treated with mesalazine (para-aminosalicylic acid) 2 g/day in combination with hydroxyurea.

The patient was closely monitored during follow-up consultations every 15 days, later transitioning to monthly visits. Clinical examination revealed an improvement in gastrointestinal symptoms, including regression of abdominal pain and diarrheal episodes, improvement in general condition, and restoration of appetite. The evolution was favorably marked by a drop in the platelet count to 286,000 elements/mm^3 in December 2021. The patient did not experience any adverse effects from the treatment, and no therapeutic adjustments were required.

3. Discussion

The incidence of Essential thrombocythemia is estimated at 1.2 to 3.0 per 100,000 population per year. The median age at diagnosis is 58 years, 67% are women [6] [7].

Essential thrombocythemia joins the group of myeloproliferative syndromes in their pathophysiology including the presence of the JAK2 tyrosine kinase mutation (JAK2V617F mutation), the discovery of which dates back to 2005. This mutation is present in 90% of polycythemia vera, 60% of essential thrombocythemia, and approximately 50% of myelofibrosis primary [8] [9].

The clinical presentation is variable, the symptoms include Vaso-occlusive accidents, hemorrhages and Arterial or venous thrombosis. The splenomegaly is rare. Sometimes, patients are asymptomatic. Moreover, the disease can be diagnosed at a stage of complications (transformation to acute leukemia or rare and late myelodysplasia) [1].

The exclusion of secondary or reactive causes of thrombocytosis (ferritin level and erythrocyte sedimentation rate) is primordial as well as infections or cancers [6].

Most patients with essential thrombocythemia harbor a mutation in one of three genes: JAK2 V617F (in 60%), CALR (in 20%), or MPL in (3%). The Confirmation of the diagnosis requires a bone marrow biopsy [6] [7].

The therapeutic management is based on thrombotic risk factors. Patients classified as low risk (<40 years old, no history of thrombosis, and platelet < 1500 G/L) do not require any treatment. However, patients classified as high-risk (40 and 60 years or >60 years or history of thrombosis or platelets > 1500 G/l) require treatment based on the platelet count (cytoreductive, interferon, hydroxyurea, and anagrelide) [1].

Ulcerative colitis is a chronic inflammatory disease affecting the colon, and its incidence is rising worldwide. The pathogenesis is multifactorial, involving genetic predisposition, epithelial barrier defects, dysregulated immune responses, and environmental factors. Patients with ulcerative colitis have mucosal inflammation starting in the rectum that can extend continuously to proximal segments of the colon.

Ulcerative colitis usually presents with bloody diarrhea. The diagnosis is based on the colonoscopy and histological findings. Management aims to induce and maintain remission, which is defined as the resolution of symptoms and endoscopic healing. Treatments for ulcerative colitis include 5-aminosalicylic acid drugs, steroids, and immunosuppressants. Some patients can require colectomy for medically refractory disease or to treat colonic neoplasia [2].

An association between hematological cancers and inflammatory bowel disease (IBD) has previously been suggested, but the risk of IBD in patients with myeloproliferative neoplasms (MPNs) is unknown. The association between ulcerative colitis and essential thrombocythemia is rare and the literature is poor in terms of this association.

A cohort study conducted in 2020 that investigated the risk of Inflammatory Bowel Disease (comprising ulcerative colitis UC and Crohn's disease CD) in Patients with Chronic Myeloproliferative Neoplasms (including essential thrombocythemia, polycythemia vera, and myelofibrosis). According to the study, the results that

concerned our association are as follows: from a total of 40 patients followed for essential thrombocythemia, 18 of them developed ulcerative colitis [5] [10].

Chronic inflammation seems to be involved in the pathophysiology of both IBD and MPNs and it contributes significantly to the symptom burden as well [11]-[15]. Inflammatory bowel disease (IBD) is an immune-mediated disorder leading to chronic inflammation of the gastrointestinal tract. Cancer is a major health problem and a common cause of death worldwide. The relationship between immune-mediated disease and cancer, however, is complex. Immune-mediated diseases often result from dysregulated and hyperactive immune responses, and active immune responses may be protective against cancer. On the other hand, immune-mediated diseases usually cause chronic inflammation, which may promote cancer development. Therefore, immune-mediated diseases may be ambivalent in suppressing and promoting cancers. In addition, immunomodulators used for the treatment of immune-mediated diseases may also affect cancer risk [3].

Moreover, shared genetic predisposition has been demonstrated in MPN and inflammatory diseases, including IBD [16]-[18]. Patients with IBD frequently have hematologic abnormalities suggestive of JAK2 mutated MPNs but are traditionally classified as reactive processes. Haplotype 46/1 is a well-characterized genetic predisposition, common to both inflammatory bowel disease (IBD) and myeloproliferative neoplasms (MPN) [4].

There are no studies in the literature that specify a particular therapeutic to patients presenting this association, in our case, the patient was treated by cytoreductive treatment based on hydroxyurea combined with Mesalazine, and she didn't develop any complications such as acute leukemia or myelodysplasia during her follow up.

4. Conclusion

Our case is special because of the rare association between essential thrombocythemia and Hemorrhagic colitis. We have to increase the number of patients included and then carry out a case-control study to analyze better and identify the links between these two pathologies as well as well-adapted therapeutic management.

Informed Consent

The patient provided clear and well-informed written consent for the publication of the case.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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