MEDWIN PUBLISHERS
Committed to Create Value for Researchers

Haematology International Journal
ISSN: 2578-501X

Pulmonary Embolism Revealing Acutisation of Vaquez Disease: A Case Report at the Haematology Department of the National Blood Transfusion Center in Bangui (Central African Republic)

Saint-Cyr Sylvestre PDV[1,2]*, Youssouf O[2], John O[2] and Serge Magloire CP[3]

[1]Clinical Hematology Department of National Blood Transfusion Center, Bangui
[2]Internal Medicine Department, Teaching Hospital of Communautaire, Bangui
[3]Hepato-gastro-enterology department, Teaching Hospital of Amitié Sino-Centrafricaine, Bangui

Case Report
Volume 7 Issue 2
Received Date: October 19, 2023
Published Date: November 08, 2023
DOI: 10.23880/hij-16000219

***Corresponding author:** Packo Dieu-le-veut Saint-Cyr Sylvestre, clinical haematology department, national Blood transfusion Center, Bangui, Tel: 00236 72 42 12 16; Email: stcyrpacko@yahoo.fr

Abstract

Introduction: Primary polycythemia belongs to the group of myeloproliferatifs disorders whose natural course is marked by thrombohemorrhagic events in the short term and transformation into acute leukemia in the long term. Primary polycythemia is rarely seen in our department. It usually precedes a myelodysplastic phase. Arterial thrombosis as a revealing mode is exceptional.

Case report: We report the case of a 78-year-old woman diagnosed with polycythemia vera in 2018, with signs of hyperviscosity in the setting of arterial hypertension treated with hydryuree. The appearance of unexplained dyspnea in 2023 led to the diagnosis of pulmonary embolism. The follow-up blood count unexpectedly revealed a bicytopenia consisting of severe anemia at 5g/dl and thrombocytopenia at 88,000/mm3. The myelogram was consistent with AML. The evolution was rapidly fatal in our context. The interest of this work lies in the rarity of this AML secondary to polycythemia vera, but also in its exceptional mode of manifestation, making management ambiguous due to treatment of a pulmonary embolism in a context of thrombocytopenia.

Conclusion: This is the first case of its kind to be described in our department.

Keywords: Acute Leukemia; Acuitization; Vacquez Disease

Abbreviations: MPS: Myeloproliferative Syndrome.

Introduction

Polycythemia vera or vaquez disease is a myeloproliferative syndrome (MPS) characterized by proliferation of the erythrocyte lineage. It is accompanied by a mutation in the gene coding for the JAK2 (Janus kinase 2) protein tyrosine kinase, called JAK2 V617F, found in 90% of cases [1,2]. Short-term complications are dominated by arterial thrombosis and, secondarily, hemorrhagic syndrome. One of the complications of SMP is transformation into acute myeloblastic leukemia (AML), which occurs in 5% to 15% of cases at the heart of Vaquez disease [1]. AML is manifested

by an altered general state, infectious or haemorrhagic syndrome, the consequences of pancytopenia. Rarely, they are accompanied by arterial thrombosis [3]. The authors report a case of transformation of vaquez disease into AML after 7 years of treatment with hydroxyurea, revealed by thrombosis of the pulmonary arteries, the prognosis of which was dismal and rapidly fatal. This is the first description of its kind in our department, prompting the presentation of this case in order to present the therapeutic and evolutionary particularity.

Case Report

Mrs B.A, aged 78, consulted the haematology department in March 2016 with a headache and vertigo. Her comorbidity was hypertension stabilized on ibarsatan+Hydrochlorothiazide. The clinical examination concludes to a hyperviscosity syndrome. There was no splenomegaly or hepatomegaly. The blood count showed 10100 white blood cells/mm^3 including 3900 neutrophils/mm^3, 17.6 g/dl hemoglobin, 59% hematocrit and 203000 platelets/mm^3. Polycythaemia was suspected. Molecular examination confirmed the presence of a JAK2 V617F mutation, leading to the diagnosis of polyglobulia vaquez.

Treatment with hydroxyurea 1000 mg/day combined with ASPEGIC 100 mg/day was initiated. This treatment stabilized the hematocrit level and had a favorable effect on functional signs. In February 2023, a sudden onset of progressively worsening exertional dyspnea associated with grade 3 physical asthenia was observed. Clinical examination revealed crackling rales in the lung fields. Cardiac ultrasound revealed right ventricular hypertrophy associated with signs of pulmonary artery hypertension suggestive of pulmonary embolism. Pre-therapeutic workup revealed a bicytopenia consisting of profound anemia at 5g/dl and thrombocytopenia at 88,000/mm^3.

The patient received a transfusion of packed red blood cells, with a stationary course marked by transfusion inefficiency. A peripheral blood smear showed 11% peripheral blastosis, prompting a myelogram which confirmed acute myeloid leukemia. The evolution was rapidly fatal within 72 h of diagnosis in a cerebral hemorrhagic syndrome picture.

Discussion

Described for the first time by William Dameshek in 1951, Vaquez disease (or primary polyglobulia, or Vaquez polyglobulia, or polycythemia vera in English) is a hematological malignancy belonging to the group of myeloproliferative syndromes. It is characterized by proliferation predominantly of the erythroblastic lineage [1,4]. Mutation of the JAK2 (Janus kinase 2) tyrosine kinase, leading to substitution of a valine by a phenylalanine at position 617, is the anomaly found in 95% of this disease [5]. Its incidence is 20 cases per 100,000 people per year and mainly affects adults aged 60 to 80, although it may occur before the age of 40 [6]. Polycythemia of Vaquez is often revealed by a haemogram showing an increase in erythrocyte parameters such as haemoglobin and haematocrit. Clinical signs may also include cutaneous erythrosis and neurological disorders linked to hyperviscosity.

The latter is manifested by headache, dizziness, paresthesia, sometimes deficit signs [7]. In our patient, headache and dizziness were the main inaugural symptoms. Diagnostically, the WHO 2016 criteria, which associate increased globular mass with the presence of a JAK2-V617F mutation, are retained for the diagnosis [8]. In our series, initial hemoglobin was 19 g/dl and molecular biology revealed a JAK2-V617F mutation. Therapeutically, treatment with hydroxyurea was rapidly initiated, given the comorbidity with arterial hypertension [9].

Hemoglobin values stabilized. Acute leukemia can complicate vacquez disease in the long term. This natural evolution is observed in 5 to 10% of cases [1]. On the other hand, the use of Hydroxy-Urea has been incriminated as one of the causes of this long-term complication [10]. In a prospective study of 150 patients, Rain JD, et al. found an actuarial risk of leukemic transformation of 10% at year 13 [10]. In contrast, this complication is observed in our series from the 7^{th} year of hydroxyurea treatment.

The revealing mode of this complication was the exception in our series. According to Brinson R et al. the mechanism is related to either leukostasis or intravascular activation of coagulation (IVAD) [10]. The use of anticoagulants was controversial due to associated severe thrombocytopenia. This makes our patient's prognosis bleak, especially in our context where chemotherapy of such a condition is not possible given our technical platform.

Conclusion

We report an unusual case in our department of acute leukemia secondary to VD, with a peculiar revealing and therapeutic mode. This should prompt practitioners to reinforce follow-up of VD with clinical examinations and a close blood count. An in-depth study will be necessary to further understand the mechanism of this association.

Authors' contributions: Dr PACKO Dieu-le-veut Saint-Cyr sylvestre is the principal investigator of this work. Dr Oumarou youssouf and Dr Onambélé john followed this patient in the internal service. Pr Camengo Police Serge Magloire provided scientific correction and guidance.

References

1. Valérie Ugo. Maladie de Vaquez (2006) Rev Pract 56: 1941-1948.

2. Staerk J, Kallin A, Royer Y, Diaconu CC, Dusa A, et al. (2007) JAK2, the JAK2 V617F mutant and cytokine receptors. Pathol Biol 55(2): 88-91.

3. Rufera A, Balabanovb S, Goedeb JS, de Vaquez M (2013) Forum Med Suisse 13(49): 1003-1009.

4. Scott LM, Tong W, Levine RL, Scott MA, Beer PA, et al. (2007) JAK2 exon 12 mutations in polycythemia vera and idiopathic erythrocytosis. N Engl J Med 356(5): 459-468.

5. Quintas Cardama A, Kantarjian H, Manshouri T, Luthra R, Estrov Z, et al. (2009) Pegylated interferon alfa2a yields high rates of hematologic and molecular response in patients with advanced essential thrombocythemia and polycythemia vera. J Clin Oncol 27(32): 5418-5424.

6. Landolfi R, Di Gennaro L, Barbui T, De Stefano V, Finazzi G, et al. (2007)Leukocytosis as a major thrombotic risk factor in patients with polycythemia vera. Blood 109(6): 2446-2452.

7. Tulliez M. Polyglobulie de Vaquez (1997) Rev fr labo 296.

8. Teffri A, Thiele J, Vardiman JW (2009) The 2008 Who classification system for Myeloprolférative neoplasms. Cancer 115(17): 3842-3847.

9. Chomel JC, Sorel N, Mayeur Rousse C, Tuhran AC (2009) Les syndromes myeloproliferatifs. Rev L 24: 69-85.

10. Brinson RR, Garcia GJ (1984) Acute lymphoblastic leukemia manifested as aortic occlusion. Am J Med 77(2): 385-387.