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Chronic Splenomegaly in a young Congolese man revealing Chronic Myeloid Leukemia: a clinical trap for clinicians in resource-limited countries.

Splénomégalie chronique chez un jeune Congolais révélant une leucémie myéloïde chronique : un piège clinique pour les cliniciens des pays à ressources limitées

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Abstract

Chronic myeloid leukemia (CML) is one of the most common chronic leukemias. Its diagnosis and treatment pose a significant challenge in sub-Saharan African countries due to the presence of several confusing pathologies and the lack of diagnostic and therapeutic tools, especially when it occurs in young patients. In the present report, a 34-year-old Congolese man was diagnosed with BCR-ABL transcript CML and fortunate enough treated with Imatinib mesylate. Here we describe his clinical picture, his treatment, and his clinical course.

Keywords: Chronic Splenomegaly, Chronic myeloid leukemia, BCR-ABL, Tyrosine kinase inhibitors, case report

Résumé

La leucémie myéloïde chronique (LMC) est l'une des leucémies chroniques les plus fréquentes. Son diagnostic et son traitement représentent un défi majeur en Afrique subsaharienne en raison de la présence de plusieurs pathologies pouvant prêter à confusion et du manque d'outils diagnostiques et thérapeutiques, notamment chez les patients jeunes. Dans le présent rapport, nous décrivons le cas d'un Congolais de 34 ans atteint de LMC avec mutation BCR-ABL, traité avec succès par imatinib mésylate. Nous présentons ici son tableau clinique, son traitement et son évolution

Mots-clés : Splénomégalie chronique, leucémie myéloïde chronique, BCR-ABL, inhibiteurs de la tyrosine kinase, rapport de cas

Introduction

Chronic Myeloid leukemia (CML) accounts for over 30 % of all leukemias, and is by far the most common type in Africa [1,2]. Its prevalence in the Democratic Republic of the Congo is poorly known, due to a lack of diagnosis tools; hence its rare description. The clinical manifestations of CML combine altered general condition of varying degrees with variable physical signs, including splenomegaly in two-third of cases. The spleen is involved in various pathological processes through its multiple immune, hematopoietic, macrophagic and vascular filtering functions. In addition, the African and tropical context of splenomegaly offers a wide spectrum of etiological diagnoses, including hemolytic syndrome, infections, portal hypertension, hematological malignancies and lymphomas. However, the limited diagnostic skills of peripheral practitioners and poor integration of blood count data lead to diagnostic and therapeutic erraticism. This leads to poor detection of malignant blood disorders, particularly CML, which is often confused with tropical splenomegaly. Splenectomies are then carried out, resulting in significant morbidity and mortality [3]. The definitive diagnosis of CML consists of evidence of significant leukocytosis with an increase in myeloid precursors without a leukemic gap, associated with the detection of a Philadelphia chromosome (translocation t9,22) and a BCR-ABL transcript on the myelogram and/or peripheral blood [4]. This poses a significant diagnostic challenge due to the lack of qualified personnel and, above all, adequate technical facilities, in this case enabling cytogenetic testing to be carried out. Treatment, which is often difficult to access in African countries, has historically involved allogeneic bone marrow transplantation, cytotoxic agents (Hydroxyurea and busulfan), then interferon alpha combined with cytarabine arabinoside, and finally, more recently, tyrosine kinase inhibitors [5]. We describe the diagnostic and evolutionary aspects of a young patient in whom splenomegaly was the main presenting sign.

Observation

A 34-year-old man, Congolese teacher at a secondary school, newly married, with no previous history consulted for abdominal enlargement and pain in the left hypochondrium, present for 6 months. He did not report any fever but had occasional joint pain that appeared to be inflammatory in nature.

Clinical findings: Physical examination revealed a patient in relatively good condition with vital signs within normal limits. The only notable finding on physical examination was an enlarged spleen, 17 cm below the costal margin, with an irregular, non-tender surface. There was no peripheral polyadenopathy, no hepatomegaly, and no abdominal collateral circulation. The joint examination did not reveal any signs of arthritis, and the skin and integument examination found no signs of petechiae or other hemorrhagic signs.

Diagnostic assessment

A complete blood count showed leukocytosis at 195.7 G/L, predominantly neutrophilic at 154.2 G/L, MID (basophils + eosinophils + monocytes) at 25 G/L; normocytic anemia with hemoglobin at 11.5 g/dL and thrombocythemia at 616 G/L. The Chronic myeloid leukemia was then suspected and RQ-PCR on peripheral blood was performed and the EUTOS score was calculated at 73.5. Negative HBsAg, anti-HCV antibody and HIV determination ruled out hepatitis B and C, as well as active HIV infection. A thick blood smear with staining and parasite density returned negative. CRP was negative and a sedimentation rate of 10 mm in the first hour ruled out an ongoing inflammatory syndrome. Transaminases were normal at 11.1 IU/L for AST (8-30) and 23.7 IU/L for ALT (6-30), and a normal ultrasonographic appearance of the liver with no signs of portal hypertension was observed. In addition, it has been observed the absence of deep abdominal adenopathy made lymphoma less likely. The absence of hemoglobinuria on urine dipstick ruled out vascular hemolysis. There was hyperuricemia at 7.6 mg/dL, hyperkalemia at 6.51 mmol/L (3.53-5.50), hypercalcemia at 1.42 mmol/L (1.11 - 1,41). Renal function was normal, with creatinine at 1.23 mg/dL and GFR estimated at 86.88 ml/min/1.73 m2. A peripheral blood smear revealed blasts. The patient was fortunate enough to have a PCR test performed on a sample sent to Kinshasa. The QR-PCR performed on peripheral blood confirmed the diagnosis of CML with the detection of a BCR-ABL transcript estimated at 38.6%.

Therapeutic interventions: Initial treatment consisted of HYDROXYUREA 1g twice daily, then reduced to 500 mg twice daily and continued for 3 months. Electrolyte disorders were progressively managed and therapy with IMATINIB was subsequently started at a regimen of 400 mg daily from the end of the third month.

Follow-up and outcome of interventions: Progression was favorable, with normalization of spleen size and blood count parameters at 4 months on IMATINIB. Leukocytes fell to 4.5 G/L, neutrophils to 1.7 G/L, MID to 0.3 G/L, platelets to 110 G/L and hemoglobin normalized to 14.5 g/dL. There was no hepatic cytolysis, rather minimal transient renal function impairment with creatinine at 2.1 mg/dl (GFR = 46.58 ml/min) while taking IMATINIB. A few clinical adverse events, such as petechiae on

HYDROXYUREA and a flaming subungual hemorrhage, were described but were not extremely severe. RQ PCR performed at 6 months of treatment detected the BCR-ABL transcript at a rate of 0.53% (IS), i.e. MR = 2.28 (less than 4). Clinical course: The patient showed improvement after only a few months under IMATINIB. He noted the "absence" of asthenia and observed for the first time a reduction in abdominal swelling. He remained confident of a "lasting/permanent" "recovery."

Discussion

CML predominantly affects men of all ages, with racial and regional variability in peak incidence. Our patient was 34-year-old at diagnosis, which is close to the 38.5 years mean age at diagnosis in central Africa [6]. The patient's general condition deteriorates only slightly, with moderate weight loss due to anorexia caused by gastric discomfort secondary to the mechanical effect of splenomegaly, which is consistent with our observation [7]. Splenomegaly is the main clinical sign at diagnosis and has been described in over 60 % of cases [8]. Depending on the course of the disease, polyadenopathy may be associated with the accelerated or blast crisis phase. Similar to our patient, leukocytosis is always present, predominantly neutrophils, and may be associated with normochromic normocytic anemia and thrombocythemia or thrombocytopenia. In fact, the BCR-ABL gene resulting from translocation 9.22 codes for a protein kinase responsible for excessive proliferation of medullary stem cells and high differentiation capacity, resulting in leukocytosis and myelemia. Diagnosis was made by quantitative measurement of the BCR-ABL transcript using RT-PCR on peripheral blood. This method may be sufficient to establish a diagnosis to a certain extent. In fact, this test is part of a series of diagnostic investigations indicated in CML, including myelogram, cytogenetics, karyotype and FISH. HYDROXYUREA was started while awaiting the availability of IMATINIB therapy. That first treatment has historically been well known for its myelosuppressive effects in all myeloproliferative syndromes in general and chronic myeloid leukemia in particular. Hydroxyurea based treatment is known to be is insufficient, marked by a temporary regression of splenomegaly and leukocytosis as in our observation. Hence the switch to IMATINIB with favorable prognosis as proposed in the literature [9]. In our patient, IMATINIB was initiated after 3 months' treatment with HYDROXYUREA. Under this treatment, splenomegaly decreased significantly, leukocytes normalized and the ACR-ABL transcript was controlled at 0.53 % at 6 months, assessing a favorable clinical, hematological and molecular response. Adverse effects such as petechiae, subungual hemorrhages during flare-ups while on HYDROXYUREA, minimal hepatic cytolysis involving ALT, and slight deterioration of renal function during treatment with IMATINIB have been observed. They often progress favorably, with no significant impact on prognosis. Moreover, this prognosis was basically good, judging by the EUTOS score of 73.5 (below 77) prior to initiation of IMATINIB. In theory, treatment with IMATINIB should be continued for at least 3 years until a major molecular response is achieved, as evidenced by a negative RQ-PCR (> RM5) and stable for at least 2 years [9]. In fact, the expected targets for molecular response to treatment are < 10% transcript, then 1 % and finally virtually zero at 3 months, 6 months and over 12 months of treatment respectively. Transcript levels < 0.01% determine major response to treatment [9,10]. Our patient, who achieved a rate of 0.53% after 6 months of treatment, therefore responded very well to the treatment in line with the objectives mentioned above.

Diagnosis of hematological malignancies mainly CML remains a challenge in African context. Splenomegaly, which is one of its main clinical manifestations, seems to make it more difficult to diagnose, given the wide range of possible etiological diagnoses, which include various tropical parasitic diseases such as malaria. In addition, the scarcity of hematologists reduces the chances of diagnostic confirmation. Difficult access to the latest generation of treatment with tyrosine kinase inhibitors makes patients' prognosis bleak. The use of generic molecules, even first-generation ones with anti-tyrosine kinase activity, could be a good alternative.

This presentation shows the importance of training in dealing with these conditions and the possibility of providing effective care in countries with limited resources.

Competing interests: None

Author's contributions

JKK: Management of the patient, writing, original draft. DB: Facilitation for QR-PCR, JM: Basic hematological explorations. TAS: writing, review and editing. All authors read and approved the final version of this manuscript

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References

1. Nwannadi I, Alao O, Bazuaye G, Nwagu M, Borke M. Clinical and Laboratory Characteristics of Patients with Leukaemia in South-South Nigeria. The Internet Journal of Oncology. 2009 Volume 7 Number 2.
2. Parkin DM, Bray F, Ferlay J, Pisani P. Global cancer statistics, 2002. CA Cancer J Clin. 2005 Mar-Apr;55(2):74-108. doi: 10.3322/canjclin.55.2.74.
3. Bonnet S, Guédon A, Ribeil JA, Suarez F, Tamburini J, Gaujoux S. Indications and outcome of splenectomy in hematologic disease. J Visc Surg. 2017 Dec;154(6):421-429. doi: 10.1016/j.jviscsurg.2017.06.011.
4. Goldman JM. Chronic myeloid leukemia: a historical perspective. Semin Hematol. 2010 Oct;47(4):302-11. doi: 10.1053/j.seminhematol.2010.07.001.
5. O'Brien SG, Guilhot F, Larson RA, Gathmann I, Baccarani M, Cervantes F, Cornelissen JJ, Fischer T, Hochhaus A, Hughes T, Lechner K, Nielsen JL, Rousselot P, Reiffers J, Saglio G, Shepherd J, Simonsson B, Gratwohl A, Goldman JM, Kantarjian H, Taylor K, Verhoef G, Bolton AE, Capdeville R, Druker BJ; IRIS Investigators. Imatinib compared with interferon and low-dose cytarabine for newly diagnosed chronic-phase chronic myeloid leukemia. N Engl J Med. 2003 Mar 13;348(11):994-1004. doi: 10.1056/NEJMoa022457.
6. Mukiibi JM, Nyirenda CM, Paul B, Adewuyi JO, Mzula EL, Malata HN. Chronic myeloid leukaemia in central Africans. East Afr Med J. 2003 Sep;80(9):470-5. doi: 10.4314/eamj.v80i9.8744.
7. Jacobs P, King HS, Dent DM. Chronic granulocytic leukaemia. A 10-year experience in the Black, Coloured and White populations of the south-western Cape Province. S Afr Med J. 1983 Jun 4;63(23):879-82.
8. Druker BJ, Guilhot F, O'Brien SG, Gathmann I, Kantarjian H, Gattermann N, Deininger MW, Silver RT, Goldman JM, Stone RM, Cervantes F, Hochhaus A, Powell BL, Gabrilove JL, Rousselot P, Reiffers J, Cornelissen JJ, Hughes T, Agis H, Fischer T, Verhoef G, Shepherd J, Saglio G, Gratwohl A, Nielsen JL, Radich JP, Simonsson B, Taylor K, Baccarani M, So C, Letvak L, Larson RA; IRIS Investigators. Five-year follow-up of patients receiving imatinib for chronic myeloid leukemia. N Engl J Med. 2006 Dec 7;355(23):2408-17. doi: 10.1056/NEJMoa062867.
9. Masarova L, Cortes JE, Patel KP, O'Brien S, Nogueras-Gonzalez GM, Konopleva M, Verstovsek S, Garcia-Manero G, Ferrajoli A, Kadia TM, Ravandi-Kashani F, Borthakur G, DellaSala S, Estrov Z, Jabbour EJ, Kantarjian HM. Long-term results of a phase 2 trial of nilotinib 400 mg twice daily in newly diagnosed patients with chronic-phase chronic myeloid leukemia. Cancer. 2020 Apr 1;126(7):1448-1459. doi: 10.1002/cncr.32623.
10. Soverini S, De Benedittis C, Mancini M, Martinelli G. Best Practices in Chronic Myeloid Leukemia Monitoring and Management. Oncologist. 2016 May;21(5):626-33. doi: 10.1634/theoncologist.2015-0337.