Unusual association case of Janus Kinase 2 Mutation and BCR-ABL transcripts : Successfull treatement with Hydroxyurea

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Background

• Mutations like JAK2 are mutually exclusive events in myeloproliferative neoplasms (MPN).

•The presence of mutations (like JAK2) is seen in BCR-ABL1-negative MPN, but its presence with Chronic myeloid leukemia (CML) is a rare finding; only a few cases have been reported .

Objective

•In this case, we describe a clinical case of patient who had both mutations (BCR-ABL and JAK2 V617F) at our hematology department of Ibn Rochd hospital

Case reports

Table 1 . Patient characterestics at diagnosis (2017)

AGE	37
SEX	Men
HISTORY	• Artériel hypertension • Chronic smoker • Occasionnel alcoolic • Amputation of the right fifty-fifty toe (2013)
Clinical Symptoms	• Fatigue • Absence of splenomegaly and hepatomegaly • Absence of lymph nod
Biological Workup	• Thrombocythemia (PL = 1,783,000/mm3) • Neutrophil leukocytes (21,300/ L) • Hemoglobin level of 12 g/L
Bone marrow	• Hypercellular • Increase number of megakaryocytes • Normal erythroid and granulocyte proliferation • Delicate reticulin fiber, absence of fibrosis • No sign of malignancy
Transcrit BCR-ABL (M-bcr) By RT-PCR quantification	0,082%
JAK 2 V617	Positive
Karyotype	46,XY

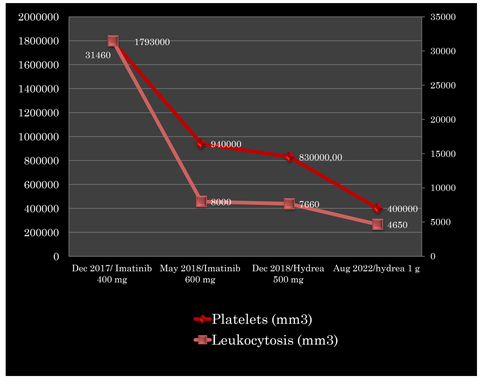


Figure 2. Treatment and evolution

Clinical Course :

➢**In the first year :**
The patient had started Imatinib 400 mg with therapeutic failure after 3 months , therafter the dose was increased to 600 mg with still no response

➢ **In the second year** : Hydroxyurea was started at 500 mg with partial hematological response, then the dose was increased to 1 g after 3 months with a good response .

➢currently in 2022, absence of BCR-ABL transcrit detection

Discussion

▪Several cases of concomitant JAK2 MPD and BCR ABL transcript have been reported. This hybride disease is more likely to be misdiagnosed because it is underdiagnosed.

▪Patients with association had a poor response to imatinib(3), which was also the case in our patient, who had a good hematological response to hydrea but a poor response to imatinib.

▪While it is unclear whether these occurrences are true associations or coincidental, the recent discovery that JAK2 may be an important therapeutic target for Ph CML .

References

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