Myelodysplastic syndrome without ring sideroblasts and with Janus kinase 2 gene mutation: An unusual case report

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INTRODUCTION

Myelodysplastic syndromes (MDS) correspond to a heterogeneous group of hematological diseases that are characterized by impaired hematopoiesis. The presence of thrombocytosis in the patients may be related to both essential thrombocytosis as MDS leading to difficulty in diagnosis. In general, patients that present thrombocytosis associated with the presence of ring sideroblasts are diagnosed as MDS, while the absence of ring sideroblasts in the patient is diagnosed as essential thrombocytosis. The case of a patient with MDS was diagnosed as essential thrombocytosis. A female patient with 78 years of age, anemia and initial diagnosis of essential thrombocytosis, was studied. The patient presented an atypical evolution of the clinical condition, characterized by thrombocytopenia and leukocytosis. Thus, an accurate study of clinical case was performed with the use histopathological, immunophenotypic, molecular and molecular cytogenetics data of the patient after her death.

METHODOLOGY

The woman patient with 78-year-old, presented macrocytic anemia and thrombocytosis, with platelet count of 812x109l. The patient presented a leukocyte count of 6.5x109/l without circulating blasts. The molecular tests revealed the presence of a JAK2 gene mutation. After 2 years of treatment, the patient presented alterations of symptoms with weight loss, epistaxis, tachycardia, fever, splenomegaly and the presence of thrombocytopenia (61x109/l) and leukocytosis (12x109/l) with circulating blasts.

The atypical evolution of the patient's clinical picture led to a more refined study of tests data after her death, that allowed a new classification of the patient's diagnosis as MDS. The patient's data during the course of the evolution of her clinical condition were compared.

Thus, a more accurate analyses was performed using immunophenothypical tests (with CD34, CD33, CD117, CD56, anti-HLA-DR (dim) and CD15 markers) and histopathological tests, such as hematoxilin-eosin staining and reticulin staining,that evaluated the bone marrow cellularity.

The ocurrence of chromosomal abnormalities was verified too through conventional cytogenetic (Trypsin- Giemsa banding), and chromosome painting. In addition, the data were analyzed to evaluate the presence of ringed sideroblasts in patient's samples through of Perls' staining.

RESULTS

The case study showed that the patient presented a Janus kinase 2 gene (JAK2) mutation. In addition, the data indicate that in patients with MDS, the mutation in the JAK-2 gene is usually associated to presence of ring sideroblasts, which were not found in the present case. The study of molecular cytogenetics data showed alterations on chromosome 3q and monosomy on chromosome 7, that confirmed the diagnosis of MDS.

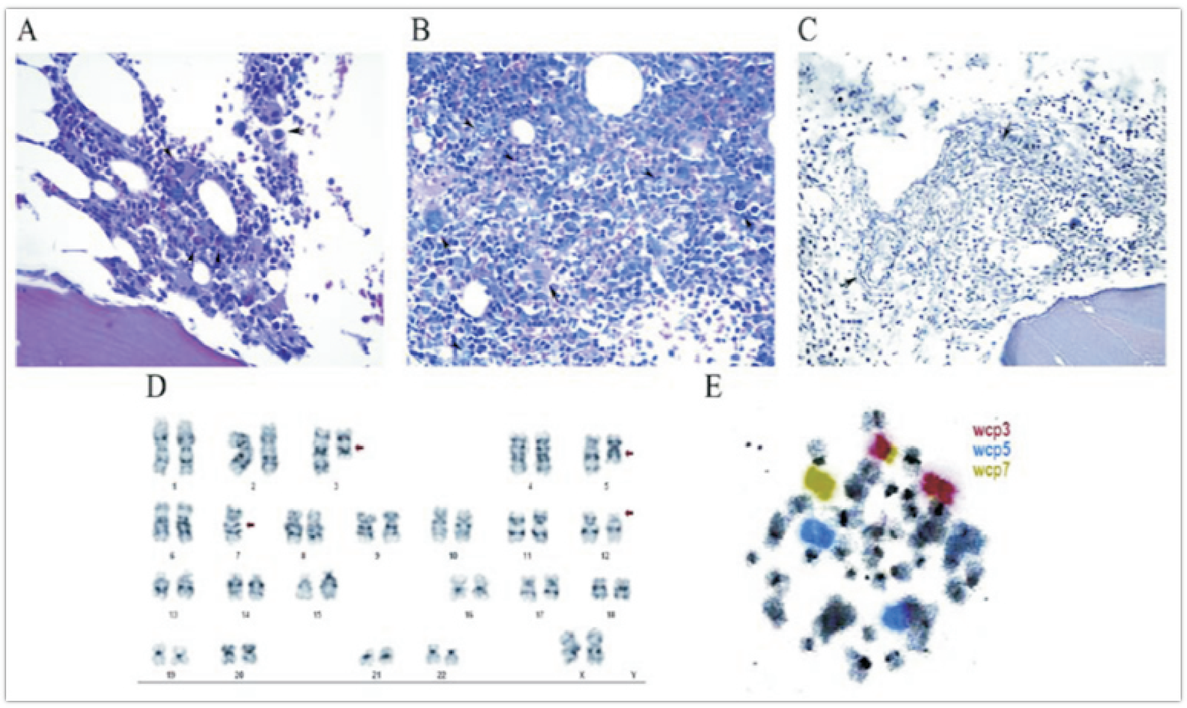

Figure 1. Histopathology and cytogenetic data. (A) Bone marrow biopsy initial (hematoxylin and eosin (H & E) staining) x400. **(B)** Bone marrow biopsy in evolution (H & E) staining) x400. **(C)** Bone marrow biopsy in evolution (reticulin staining) x400. **(D)** Trypsin-Giemsa banding. **(E)** Chromosome painting.

CONCLUSION

The correct diagnosis and treatment of myelodysplasia depend, in together, of the analysis of clinical, immunophenotypic, histopathological, molecular, cytogenetic and molecular cytogenetic data

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