Myelodisplastic syndromes: case presentation

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- Patient profile: women, 74 years old, retired, the urban environment

Anamnesis: she was presented to the doctor for little effort dyspnea, fatigue, dizziness

- Family and personal physiological and pathological history:B-2 A-6, menopause, discopathy lumbar, perineal old tear, old myocardial infarction (1993), hypertension, congestive failure cardiac class. NYHA III, cholecystectomy, right basal pneumonia (4 episodes , the last in august 2008), aortic disease
- Living / work and associated risk factors: not important

History of present illness:

- Insidious onset disease, discovered anemia (Hb 9.76 g / dl) in 2007, without treatment, without further investigation
- Between august 2008 and march 2009 the patient was hospitalized 3 times in Fundeni Medical Clinic for right basal pneumonia, during which it finds anemia (Hb 7.2 g / dl, MCV 127fl), sideremie low, but not revealed a source of bleeding in the digestive tract or genital investigations . She started treatment with iron and folic acid.
- In March 2009 she was admitted in Fundeni Clinical Hematology with pallor, mitro-aortic systolic murmur, liver 2 cm under rebord, S at rebord, KI 80%

The investigations conducted showed: macrocytic anemia, sideremie, dosage of vit.B12, folate- normal, ferritin level increased, negative Coombs test, increased serum erythropoietin level (1700mUI/ml)

- Bone marrow examination: mbl 1%, agranulare hypogranular neutrophils (but <10% cells in the granulocytic series), erythroblast series quantitatively normal (22%), predominantly normoblastic, most policromatofile, rare erythroblasts in mitosis,
- MH present, moderately increased quantity, 54% sideroblasts, 36% ringed sideroblasts
- Bone marrow biopsy: increased cellularity (80/20), dysplastic red cells, dysplastic megakaryocytes
- BM cytogenetic examination (20 metaphases): complex karyotype, over 3 clonal chromosomal abnormalities.

The positive diagnosis, according to WHO criteria was for Refractory Anemia with Ringed Sideroblasti, intermediate-1 IPSS and WHO based prognostics scoring system (WPSS) - 3.

Differential diagnosis was made mainly with: vitamin B12 and folic acid deficiency, Refractory Cytopenia with Multilineage Dysplasia, MDS with del (5q).

Therapeutic targets: the older a patient with multiple heart disease, we aimed to achieve a satisfactory quality of life. Treatment was performed, as LeukemieiNet indications with recombinant human EPO (although according to score Scandinavian-American rate of response is 23% in this patient), substitution and iron chelator treatment started as early to avoid adverse reactions of iron overload.

Clinical and paraclinical follow up in dynamics: monthly blood counts, biochemistry, sideremie were followed and at 6 months: blood counts, biochemistry, ferritin, bone marrow examination, cytogenetic examination, echocardiography (FE), ophthalmology.
The percentage of blasts in bone marrow was kept below 5%. Cytogenetic examination showed chromosomal instability with complex cytogenetic abnormalities,.

The patient required transfusion therapy to 2-3 months in the first 20 months, then monthly. Exjade treatment was started early at a ferritin level of 1030ig/dl. Although the response to erythropoietin was partially we obtained a satisfactory quality of life, with a lower transfusion need. Next will need to carry out periodic bone marrow and cytogenetic examinations at regular intervals to detect early disease progression.