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A CASE REPORT ON MYELODYSPLASTIC SYNDROME WITH SEVERE ANEMIA

General Medicine

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ABSTRACT

Myelodysplastic syndrome (MDS) is a hematological malignancy characterized by ineffective hematopoiesis leading to peripheral blood cytopenia, hypercellular bone marrow, dysplasia and increased incidence of progression to Acute Myeloid Leukemia (AML). It commonly presents asymptomatically or with symptomatic cytopenia. Here, we present a rare case of 61-year-old women with Myelodysplastic anemia with refractory anemia who presented with fever, breathlessness and persistent cough. Pathological analysis of the bone marrow aspiration and biopsy revealed hypercellular marrow with megaloblastic erythroid and megakaryotic hyperplasia. Diagnosis was confirmed based on myelodysplastic syndrome panel by FISH and genetic profiling.

KEYWORDS

Myelodysplastic syndrome, refractory anemia, allogenic stem cell transplantation

INTRODUCTION

Myelodysplastic syndrome is a type of cancer in which the bone marrow does not make healthy blood cells (RBCs, WBCs and platelets) and there is presence of abnormal cells in the blood/bone marrow. When there are less amount of healthy blood cells, infection, anemia or bleeding may occur. Sometimes MDS may progress to Acute Myeloid Leukemia (AML). Patients are usually asymptomatic. Typical disease manifestation includes fatigue and weakness from anemia and infections from neutropenia.[1] The diagnosis of MDS is based on the findings of quantitative changes in one or more peripheral blood/bone marrow elements. (RBC, platelets, granulocytes), morphological evidence of significant dysplasia upon visual inspection of peripheral blood smears, bone marrow aspirate or bone marrow biopsy and blast forms accounting for less than 20% of the total nucleated cells of the bone marrow aspirate.

CASE STUDY

A 61-year-old female patient reported to the outpatient department with eight-month history of fatigue, persistent cough and recurring fever. Patient was newly diagnosed with Type 2 Diabetes Mellitus and was on Metformin 500 mg once daily for the same since a month. She then showed her previous laboratory reports and Complete blood count showed Pancytopenia; severe anemia, thrombocytopenia and leucopenia. Hemoglobin was 3.8 gm/dl, packed call volume 16%, platelets 81,000/mm3 and white blood cells was 3,200/mm3. She was previously treated with Erypro 40k (Epoetin alfa) 40000 units which showed poor response according to the bystander. On admission, patient's blood sample was sent for Complete Blood Count and it showed Hemoglobin levels as 6.0 gm/dl. Considering this, blood transfusion was done and Hb level raised to 7.6 gm/dl. (Table 1)

Bone marrow biopsy and aspiration was performed. Pathological analysis of the bone marrow aspiration and biopsy revealed hypercellular marrow with megaloblastic erythroid and megakaryotic hyperplasia. Acute myeloid leukemia (AML) or Myelodysplastic syndrome (MDS) was suspected and Myelodysplastic Syndrome Panel by FISH (Fluorescence In-situ Hybridization) was ordered. Reports showed abnormal diploid female karyotype

with clonal abnormalities. Interstitial deletion of region q13q33 in chromosome 5 in all 20 cells was observed.

Table 1

Complete Blood Count	Observed value		Reference Range
	On admission	After transfusion	
Hemoglobin	6.0	7.6	11.5-16.5 gm/dl
Packed Cell Volume	18.2	23.5	36-46%
RBC	$2.18^{*}10^{6}$	$3.0^{*}10^{6}$	$3.5\text{-}4.5 * 10^{6}$ /mm^3
Platelets	1.12	81,000	1.5-4.5 lakhs/mm^3
WBC	4,400	3,400	4000-11,000/mm^3
Neutrophils	32	18	40-75%
Lymphocytes	42	40	20-45%
Eosinophils	08	01	1-8%

Furthermore, genetic profiling was done to assess relevant somatic mutations and mutations were found in the following genes; CBL and RUNX1 (gene regulating tyrosine kinase), SETBP1 (protein coding gene) and ASXL1 (epigenetic regulator genes).

Patient is currently being managed with supportive measures such as blood transfusions and close monitoring.

DISCUSSION

Treatment options for myelodysplastic syndrome are supportive care, drug therapy and stem cell transportation. Allogeneic hematopoietic stem cell transplantation (HSCT) is the only curative approach for patients with MDS. Dependent upon disease status at the time of transplantation, 30–70% of patients can be expected to be cured of their disease and survive long-term.[2] Allogenic hematopoietic stem cell transplantation is often limited to younger and high-risk patients and rarely in older patients. Also due to post-transplant relapse and graft-versus-host disease (GVHD), HSCT was not considered for this patient.

Supportive care includes blood transfusions, erythropoiesis stimulating agents and antibiotic to fight infections. Anemia was one of the major concerns in this case as the hemoglobin level reached as low as 3.4 gm/dl and there was a need of repeated blood transfusions. Patients who receive many blood

cell transfusions may have tissue and organ damage caused by the buildup of extra iron which may be treated by iron chelation.[5]

Drug therapy includes Lenadomide which can be given to patients with an isolated del(5q) chromosome abnormality to lessen the need for red blood cell transfusions. Antithymocyte globulin (ATG) weakens the immune system. Azacitidine and decitabine kill cells that are dividing rapidly and may slow the progression of myelodysplastic syndromes to acute myeloid leukemia.[5]

Previously published case reports of Myelodysplastic syndrome:

Author/Year of publication	Patient's age/ Gender	Diagnosis	Symptoms	Treatment
Andrea L *et al*. 2011[3]	53 /F	MDS with autoimmunity	Shortness of breath and pleuritic chest pain	Decitabine, Methotrexate and blood transfusions
Emoke H *et al*. 2016 [3]	66 / M	MDS associated with myeloid sarcoma	Fatigue, fever and night sweats	Chemotherapy -high dose cytosine arabinoside
Wagar J *et al*. 2015 [4]	71/M	MDS with refractory thrombocytopenia	Redness and swelling in left lower limb	Correct cytopenias

CONCLUSION

MDS is a rare hematological disorder. Most patients are asymptomatic and are suspected of this condition during routine blood tests. Along with the cytopenia, diagnosis is confirmed with bone marrow biopsy, bone marrow aspirate and genetic testing. As the risk of treatment related mortality and long-term complications is associated with hematopoietic stem cell transplantation, decisions should be individualized for each patient based on their age, disease stage and presence of other conditions. Non transplant treatment options should also be considered.

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