

Age is just a Number!

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Abstract Myelodysplastic syndrome (MDS) is a group of stem cell disorders characterized by ineffective hematopoiesis resulting in cytopenias and is rarely seen in patients younger than the age of 40 years old. MDS tends to be more aggressive in the younger population. We report a case of a 26-year-old male who presented with MDS.

Keywords: Myelodysplastic syndrome, Paroxysmal nocturnal hemoglobinuria

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1. Introduction

Myelodysplastic syndrome (MDS) is a group of stem cell disorders characterized by ineffective hematopoiesis resulting in cytopenias and increased risk of acute myeloid leukemia in 1/3 of patients. [1] The median age at diagnosis is 71 years old and it is very rare in patients less than the 40 years old. [2] The estimated annual incidence of MDS is 0.5 per 100,000 persons less than age 50 and 89 per 100,000 persons greater than age 80. [3] This case presents a disease occurring at a rare age.

2. Case Presentation

A 26-year-old male with no significant past medical history presented to the emergency department for evaluation of left hip pain over the last 7 months. Further questioning revealed that his leg pain has progressed to difficulty ambulating and is associated with generalized weakness, fatigue, weight loss of nearly 50 pounds and multiple skin wounds.

Initial vitals revealed BP 106/53 mmHg, HR 123 beats per minute, RR 23 breaths per minute, temperature 97.5° F, and oxygen saturation of 86% on room air. On physical examination, he was cachectic with multiple open skin lesions on the left shoulder, left upper arm, and extending onto chest wall. The skin on the scrotum and gluteal region had multiple superficial ulcers. The right hip was mildly tender.

Laboratory analysis revealed a hemoglobin of 2.9 g/dl, MCV 81 fL, white blood cell 2.1×10^9 L, and platelet 140. Lactate 8 and potassium 2.9. Peripheral blood smear showed anisopoikilocytosis and microcytic hypochromic red blood cells. MRI of the right hip was unremarkable. The patient was started on vancomycin and piperacillin-tazobactam for severe sepsis secondary to multiple skin ulcers and abscesses. Three units of packed red blood cells were transfused. Biopsies of skin lesions were consistent with hidradenitis suppurativa. Differential diagnoses included

aplastic anemia, paroxysmal nocturnal hemoglobinuria, viral infection, Fanconi anemia, leukemia, and myelodysplastic syndrome.

During hospitalization blood cultures showed no growth, however, wound culture grew MRSA. Antibiotics were switched to clindamycin. Extensive viral work up including EBV, CMV, parvovirus, and HIV were negative. CTA of the chest showed multiple pulmonary emboli and lower extremity doppler ultrasound showed extensive deep vein thrombosis involving the left common femoral, deep femoral, and femoral proximal vein. He was initially treated with heparin which was later transitioned to enoxaparin.

Bone marrow aspirate and biopsy was consistent with mildly hypocellular bone marrow, reversed myeloid to erythroid ratio of less than 1:1 due to element of granulocytic hypoplasia with megaloblastoid changes within erythroid precursors and dyspoiesis with myeloid cells suggestive of myelodysplastic syndrome. Karyotype from bone marrow showed unbalanced translocation involving chromosomes 1 and 7 is shown in Figure 1 below. Peripheral blood flow cytometry revealed a small clone of paroxysmal nocturnal hemoglobinuria population.

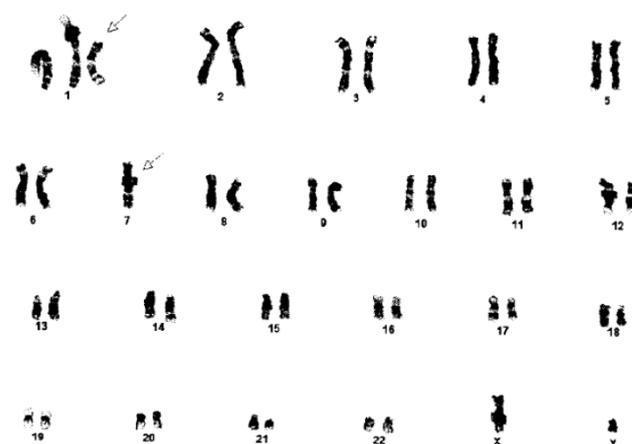


Figure 1. shows an unbalanced translocation involving chromosome 1 and chromosome 7 resulting in a net gain of 1q and a net loss of 7q

The patient was diagnosed with myelodysplastic syndrome. He was referred for hematopoietic stem cell transplantation upon discharge.

3. Discussion

Myelodysplastic syndrome should be considered in any patient who has an unexplained cytopenia. MDS can be de novo or secondary to multiple environmental insults including chemotherapy and radiation therapy. [1] The patient presented with hip pain, fatigue, leukopenia, and anemia requiring multiple blood transfusion which can all occur in patients with MDS. He was also diagnosed with biopsy-proven hidradenitis suppurative with documented cases on literature review occurring in patients with MDS.

Evidence of paroxysmal nocturnal hemoglobinuria was found on flow cytometry. This can be seen in up to 30% of patients with myelodysplastic syndrome. [4] Abnormal karyotype occurs in approximately 50% of patients with MDS and includes mutations in chromosome 7. [5] This patient karyotype showed an unbalanced translocation involving chromosomes 1 and 7. DEB clastogen assay was negative for Fanconi anemia. GATA2 gene sequencing was done given the patient's unusually young age of onset but findings showed no mutation. The patient had venous thromboembolism which can be a

complication of MDS. The definitive management of MDS is Hematopoietic stem cell transplant.

4. Conclusion

This case identifies the importance of a wide differential diagnosis, though age is often a risk factor for many diseases it does not always rule out a diagnosis. MDS is rare and tends to be more aggressive in the younger population. Allogeneic stem-cell transplantation offers younger patients the best outcomes.

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