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DIAGNOSTIC DILEMMA IN A CASE OF SYSTEMIC LUPUS ERYTHEMATOSUS

Internal Medicine

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ABSTRACT

Systemic lupus erythematosus (SLE) is a systemic autoimmune disease with multisystem involvement. Anemia is the most common hematologic abnormality in patients with SLE1. Hemoglobin E/beta-thalassemia (HbE/beta-thalassemia) is the genotype responsible for approximately one-half of all severe beta-thalassemia worldwide1. Here we report a case of severe anemia diagnosed with HbE/beta-thalassemia with sle , which is a very rare association which challenged us in therapeutic approach. In SLE patients, anemia usually results from the disease itself, but it is important to think of other coexisting conditions like thalassemia.

KEYWORDS

BACKGROUND:
Hematological manifestations are frequently seen in systemic lupus erythematosus (SLE). Anemia, leucopenia and thrombocytopenia may result from bone marrow failure or excessive peripheral cell destruction, both of which may be immune mediated[2]. Anemia is a common manifestation in patients with systemic lupus erythematosus. The most common forms of anemia, in these patients, are autoimmune hemolytic anemia, iron-deficiency anemia, anemia of chronic disease, drug-induced myelotoxicity, anemia of chronic renal failure and rarely types such as pure red cell aplasia, B12 deficiency anemia and myelofibrosis[3]. The first-line therapy for AIHA are corticosteroids, and should be slowly tapered over a time period of 6–12 months. For refractory/relapsed cases, the current sequence of second-line therapy is splenectomy, rituximab, and thereafter any of the immunosuppressive drugs (azathioprine, cyclophosphamide, cyclosporin, mycophenolate mofetil). Additional therapies are intravenous immunoglobulins, danazol, plasma-exchange, and alemtuzumab and high-dose cyclophosphamide as last resort option

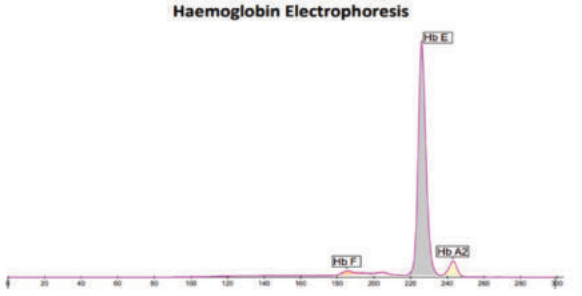
Haemoglobin E-beta thalassaemia (Hb E/β-thalassaemia) is the genotype responsible for approximately one-half of all severe beta-thalassaemia worldwide[3]. The disorder is characterized by marked clinical variability, ranging from a mild and asymptomatic anaemia to a life-threatening disorder requiring transfusions from infancy[1].Chronic blood transfusion (CBT) is essential in the management of individuals with beta-thalassemia major. However, it has major complications including iron overload that necessitates regular monitoring and treatment by long-term iron chelation therapy to protect the patients from endocrinopathies and cardiomyopathies, which can be fatal. Skeletal changes may result from the expansion of the bone marrow, and development of masses from extramedullary hematopoiesis. In spite of that, CBT causes iron overload that requires monitoring and management through long-term iron chelation therapy[5].

Case Presentation:
A 18-year-old female presented with weight loss since, loss of appetite, amenorrhea, for 6 months fever of low grade intermittent in nature for 2 months, arthralgia and oral ulcers. history of multiple blood transfusions in the past. On examination patient had severe pallor, mild icterus, without any signs of clubbing, cyanosis, or lymphadenopathy. respiratory examination revealed bilateral infrascapular diminished sounds.

Initial laboratory investigations are RED BLOOD CELL COUNT 2.58 x 106/µL (normal 3.8-4.8 x 106/µL)HAEMOGLOBIN 4.2 gm/dL (normal 12-15 gm/dL) WHITE BLOOD CELL COUNT 4.73 x 103/µL (normal 4-10 x 103/µL) MEAN CORPUSCULAR VOLUME 52.7fL (normal 80.0-100.0 fl) HEMATOCRIT 13.6% (normal 37.0-47.0 %) PLATELETS 2.82 x 103/ml(normal 150 to 400 x 103/ml RETICULOCYTE COUNT 0.9% (normal 0.5%-2.5%), SR FERRITIN 1200µg/L (normal 12-160 µg/L) TOTAL BILRUBIN 3.0 mg/dl (normal 0.3-1.2 mg/dl) DIRECT BILRUBIN 0.4 mg/dl (normal 0-0.25 mg/dl) CREATNINE 0.57 mg/dl(normal 0.6-1.2 mg/dl) UREA 30 mg/dl (normal 10-50 mg/dl), direct antiglobulin test was positive and LDH was1100 u/l.

Peripheral blood smear shows microcytic hypochromic anemia with target cells, anisopoikilocytosis, spherocytes, tear drop cells, pencil cells and helmet cells.

Bone marrow is normal reactive, Ultrasound of the abdomen was done, which was positive for mild hepatosplenomegaly. We looked for autoimmune hemolytic anemia. Due to the history of multiple blood transfusions in the past, the iron overload was evident (probably due to multiple blood transfusions) and the low mean corpuscular volume (MCV), hemoglobin electrophoresis was sent and it showed the findings in the image 1, based on that the patient was diagnosed with HbE/beta-thalassemia.



Name	%
Hb F	2.2
Hb E	92.0
Hb A2	5.8

Autoimmune workup revealed ANA HEp-2 Positive. Pattern Nuclear Speckled (AC-2,4,5) & Cytoplasmic, Intensity Nuclear Speckled (AC-2,4,5)3+ & Cytoplasmic 1+, Primary titre 1:100, End point titre Nuclear Speckled (AC-2,4,5)1:1000 & Cytoplasmic 1:100

The above investigations and peripheral smear findings suggestive of acute flare SLE with HBE/beta thalassemia in the background. she was managed as a case of SLE flare causing thrombocytopenia and anemia complicated by thalassemia, for which she was started on pulse steroid therapy for 5 days. Later, when her hemolysis symptoms resolved after 5 days transfused 3 units of packed RBC. She was also started on deferasirox 500 mg daily due to iron overload. In the next few days, the platelet count improved her Hb stabilized around 10 mg/dL. she was

discharged on oral prednisolone with a follow-up after two weeks to follow CBC. In the follow-up, her platelet count was 122 x 10^3/µL and her Hb was 9.0 mg/dL, so prednisolone was tapered with close follow-up in rheumatology and hematology clinics.

DISCUSSION:

We described a patient who has HbE/beta-thalassemia double heterozygosity, admitted with severe anaemia found to have SLE. Anemia in SLE may result from several mechanisms and more than one may be operative at any time[4]. Anemia of chronic disease is the most common, but a low hemoglobin can be caused by auto-antibodies to red cells as part of the auto-immunity, or it may be the result of impaired erythropoietin production by kidneys involved in the SLE, gastrointestinal blood loss from anti-inflammatory therapy, increased red cell destruction from hypersplenism or a drug-induced immune phenomenon[4].

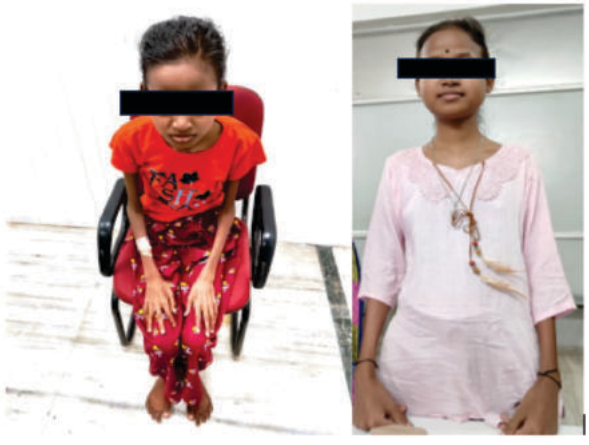
Anemia of chronic disease happens when you have an autoimmune disease or other illness lasts longer than three months and that causes inflammation[5]. Chronic inflammation can affect your body's ability to use iron needed to make enough red blood cells[5]. Anemia of chronic disease is usually mild but can be severe. It is usually normocytic, but can be microcytic. The presence of both anemia of chronic disease and dietary iron deficiency results in a more severe anemia. our patient had normal TIBC, serum ferritin which favours our diagnosis.

Patients renal function tests and ultrasound whole abdomen suggestive of normal kidney function with mild hepatosplenomegaly and bone marrow shows normal erythropoiesis. So, we can rule out anemia due to uremia and hypersplenism.

Hb E/β-thalassaemia results from co-inheritance of a β-thalassaemia allele from one parent and the structural variant Haemoglobin E from the other. The association between beta-thalassemia in general and SLE is rare, and it is rarer in HbE/beta-thalassemia based on the reported case all over the world. Upon review of literature, we have found out 2 cases reported till now this is the third case[2].However, our patient does not show any signs of hemolysis which is not the only reason that might explain the severe anemia.

CONCLUSION:

The association between beta-thalassemia in general and SLE is rare, and it is rarer in HbE/beta-thalassemia and SLE based on the reported case all over the world. More severe symptoms can present in concomitant disease. This warrants early suspicion and diagnosis of other diseases that might worsen the primary disease. Here the investigations we are showing are favouring multiple differential diagnosis causing anemia. Hence, the diagnostic dilemma.



Before **After**

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