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Case Report

Internal Medicine Section

Association of Systemic Lupus Erythematosus and Beta Thalassaemia Trait- A Case Report

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ABSTRACT

Systemic Lupus Erythematosus (SLE) is a multisystem chronic inflammatory disease of autoimmune aetiology. It has a predilection for female gender and presence of photosensitive rash over the sun exposed area gives a clue to the diagnosis. Diagnosis in a male patient with atypical manifestations is unusual and difficult. A 25-year-old male presented with fever, fatigue, vomiting, abdominal pain and loss of weight. He had sustained injury on his right arm following which he developed abscess at the trauma site and severe anaemia. Further evaluation revealed pancytopenia and peritonitis. Though peritonitis is rare in SLE, it was considered in the differential diagnosis after ruling out bacterial and tubercular peritonitis. Positive anti-dsDNA and antiSm antibodies confirmed the diagnosis. While evaluating for microcytic anaemia it was found that iron studies were normal and A2 fraction was raised in haemoglobin electrophoresis. The symptoms and laboratory parameters improved remarkably with steroid therapy. Beta thalassaemia trait is rare in patients with SLE, but when they co-exist the manifestations can be severe. High degree of suspicion is required to diagnose SLE in male patients in absence of typical photosensitive rash. Beta thalassaemia trait often does not require any treatment except genetic counseling. However empirical treatment with iron should be avoided.

Keywords: Lupus peritonitis, Microcytic anaemia, Pancytopenia

CASE REPORT

A 25-year-old male patient presented with abdominal pain, loss of appetite, low grade fever and anaemia. He had sustained injury on the right arm with an iron rod two months back. Few days later patient developed fever, chills and pain and swelling at the injured site. He was diagnosed to have abscess at the site of injury accompanied by severe anaemia with Haemoglobin (Hb) being 5.6g%. His abscess was drained and he received three units of blood during and after the procedure. He was treated with antibiotics and was discharged after significant improvement. The patient reported back about a week later with weakness and fatigue. He was found to be anaemic (Hb=6.9g%) and was transfused with three units of blood. His condition improved and he was prescribed haematinics at the time of discharge. After a few days he developed fever with chills again, this time accompanied with anorexia and abdominal pain. He sought treatment in another hospital where he was diagnosed to have ascites and pancytopenia. He received two units of blood and antibiotics over the next few days. Subsequently patient was referred to us for evaluation. At this time patient had postprandial abdominal pain and bilious vomiting. The patient complained of significant loss of weight over last two months but this could not be corroborated as weight had not been recorded in the previous medical examinations.

At the time we examined him, the patient was emaciated, pale, had a pulse rate of 90/min, BP 90/70 mmHg, respiration rate 18/min, temperature of 99.4°F. There was no rash and no history of photosensitivity. Abdominal examination revealed diffuse mild tenderness, hepatosplenomegaly and shifting dullness. Other examination was normal.

Local examination of injury site was normal, no erythema or tenderness was noted. Provisional diagnosis of primary peritonitis of infective or autoimmune aetiology was entertained. Laboratory investigation results are given in [Table/Fig-1].

Ascitic fluid was exudative in nature and was negative for acid fast bacilli, gram staining and culture. Since bacterial and tubercular peritonitis appeared to be less likely at this stage, further testing was done to look for other causes of ascites. Serum Antinuclear antibody, antidsDNA and antiSm antibody all were highly positive. There was no history of loss of blood and stool for occult blood testing done thrice was consistently negative. In view of peripheral blood smear showing microcytosis (mean corpuscular volume 72 femtoliters (fl), normal serum iron and raised serum ferritin level, haemoglobin electrophoresis was planned to rule out haemoglobinopathies. However because of recent transfusion it was postponed to subsequent visits. He was diagnosed as Systemic Lupus Erythematosus (SLE) and was treated with oral prednisolone one milligram/kg/day after breakfast. Patient improved dramatically. Abdominal pain subsided and appetite improved. His haemoglobin, leucocyte and platelet count stabilized and showed an improving trend. Haemoglobin electrophoresis was done 2 months later and showed raised Haemoglobin A2 level.

The patient was diagnosed to have Beta thalassaemia trait with concurrent SLE. He was advised not to take iron therapy empirically for anaemia and steroid was continued. On follow-up visits patient had shown side effects of steroid like swelling of face. Azathioprine, a steroid sparing drug, was added and dose of steroid was tapered.

DISCUSSION

SLE is a multisystem disease and can involve any organ of the body concurrently or successively. Haemolytic anaemia is a manifestation of SLE but coexistence of SLE with other haemolytic anaemias has been reported rarely. Castellino et al., reported that beta thalassaemia is less common in patients with SLE than in general population [1], but the symptoms of SLE were more severe if they did co-exist [2]. Their study also reported that all the patients of SLE with beta thalassaemia trait were females. Here we present co-occurrence of SLE and beta thalassaemia trait in a male patient who presented with pancytopenia and ascites. The patient did not have any rash and patient being a male, the diagnosis was a little delayed. When ANA was found to be positive antidsDNA had been ordered. To reinforce the diagnosis of SLE anti-Sm antibody test was done, which also returned as positive.

Investigations	Result	Normal range	Investigations	Result	Normal range
Hb (g%)	4.6	13-17	Blood Urea (mg%)	36	20-40
Total leukocyte count (per mm^3)	3300	4000-11000	Serum Creatinine (mg%)	1.38	0.5-1.5
Differential count (%)			Serum sodium (mmol/L)	130	135-145
Neutrophil	75	40-70	Potassium (mmol/L)	4.1	3.5-5.5
Lymphocyte	21	20-40	Calcium (mg/dL)	9.2	9.0-11.0
Eosinophil	3	0-8	Phosphorous (mg/dL)	4.3	2.5-4.5
Monocyte	1	4-11	Serum Lactate dehydrogenase (U/L)	657	100-190
Basophil	0	0-3	Total serum bilirubin(d/L)	0.9	0.3-1.9
			AST(U/L)	27	5-40
Platelets (lakh/mm^3)	0.8	1.5-4.5	ALT(U/L)	24	5-45
ESR (mm in 1st hour)	55	0-12	Serum amylase(U/L)	2891	15-200
Malaria parasite	Negative	-	Serum lipase(U/L)	1331	73-343
Dengue serology	Negative	-	Ascitic fluid protein(g/dL)	3.81	-
Serum Iron mcg/dL	101	65-175	Ascitic fluid TLC(per cmm)	1920	-
TIBC (mcg/dL)	156	250-425	Ascitic fluid		
Transferrin saturation (%)	65	20-50	Polymorph (%)	65	-
Serum ferritin (ng/mL)	830	22-322	Lymphocyte (%)	35	-
Serum Ceruloplasmin(g/L)	0.15	0.2-0.6	Asctic fluid ADA(IU/L)	1	6.8-18.2
Serum TSH(U/mL)	2.57	0.3-5.0	Anti nuclear antibody IU	8.36	<1
Serum tTG(U/mL)	1.78	<7.0	AntidsDNA IU/mL	>400	<20
HBsAG/AntiHCV	Negative	-	Anti Smith antibody IgG U	122.58	<20
HIV	Nonreactive	-	Stool occult blood	Negative	-

[Table/Fig-1]: Laboratory investigation results.

Beta thalassaemia trait is a mild disease presenting with microcytic anaemia. Patients require blood transfusion only occasionally. Treating such patients empirically with iron may lead to iron overload and should be avoided. Paul et al., suggested that whenever the transfusion requirement increases in a patient with thalassaemia, it warrants searching for an associated cause instead of blindly transfusing the patient [3]. There are reports of Beta thalassaemia trait being associated with other autoimmune diseases like rheumatoid arthritis, asthma, nephritis, diabetes. Altinoz et al., proposed that the close proximity of haemoglobin beta chain gene locus to several important genes regulating immune system may cause the haplotypal association at 11p 15.5, of these immune regulating genes with beta globin chain gene [4].

Prevalence of serositis in SLE is about 16%. Pleuritis or pericarditis constitutes one of the ACR (American College of Rheumatology) criteria for diagnosis of SLE. But peritoneal serositis (lupus peritonitis) has rarely been reported in the literature. There are various mechanism by which ascites can occur in SLE e.g., lupus mesenteric vasculitis, hypoalbuminemia [5]. Ascites can also occur in SLE when complicated by nephrotic syndrome, cirrhosis of liver or heart failure [6]. In these situations ascitic fluid will be transudative in nature. Peritoneal inflammation leading to exudative ascites usually responds to systemic steroid and nonsteroidal anti-inflammatory drugs [7]. There is a case report of massive ascites being treated with intraperitoneal steroid when patient did not respond to oral steroid [8]. In the present case patient had responded well to oral prednisolone at a dose of 1 mg /kg body weight/day.

This patient was unusual on various counts. All the previously reported cases of SLE with beta thalassaemia traits were females. The patient had lupus peritonitis which is a rare feature of the disease. Pancytopenia was one of the initial manifestations of the disease. It is difficult to say whether his disease was triggered by initial trauma. Experimental study suggests severe tissue injury can trigger severe inflammation in lupus prone mouse [9]. Rare case report of trauma triggered SLE has also been reported in the literature [10].

CONCLUSION

SLE associated with beta thalassaemia trait is rare and rarer in males. Though the overall incidence of beta thalassaemia trait is less in patients with SLE, more severe systemic symptoms are seen in concomitant disease. Awareness of such association may help us in diagnosing more cases, particularly in those with atypical presentation.

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