Wilson’s Disease with Systemic Lupus Erythematosus

RPSP Santhakumar¹, K Gayathri¹, PK Ramalingam¹, BV Manjunath¹, N Karuppusamy², B Vetriveeran², S Selvamani², P Vishnuram², A Muruganathan³, Kumar Natarajan⁴

Abstract

Case reports of Wilson’s disease occurring in combination with SLE are rarely reported in literature. Drug induced lupus have been observed in patients taking D-penicillamine for Wilson’s disease. Here we report a case from Coimbatore Medical College hospital, who presented with fever and neuropsychiatric symptoms as the initial manifestation and found to have both SLE and Wilson’s disease on subsequent evaluation.

Introduction

Neuropsychiatric symptoms in a young patient with fever can pose a diagnostic challenge. Here, we report the case of a young lady who was diagnosed with two major diseases which were causative of her symptoms.

Case Report

A 24 years female was admitted with history of 2 episodes of generalised tonic clonic seizures. The patient had fever for the past 8 months and episodic altered behaviour for past 3 months. She had lost weight over the past 5 months period and was not able to take food due to oral cavity ulcers. She also suffered from chest pain, breathlessness and frequent loose stools. She had two abortions at 12 weeks and 20 weeks of gestation respectively, in the past year. She had no chronic illness or long-term drug intake in the past.

Clinically, she was febrile, pale and malnourished. Skin was dry and scaly with ulcers in the angle of mouth and lateral aspect of tongue (Figure 1). Breath sounds were decreased in the left infrascapular region. She had diffuse abdominal tenderness. Investigations revealed a normocytic, normochromic anemia (Hb – 3.9 gm%) with a raised ESR (1/2 hr – 52 mm, 1 hr – 105 mm) and positive rheumatoid factor. Liver function test was normal. Renal parameters were marginally elevated (urea – 54 mgdL, creatinine – 1.1 mgdL) with proteinuria (200 mg/24 hrs). Diffuse cerebral atrophic changes were seen in CT brain (Figure 2). CT X-ray and chest revealed a left lower lobe collapse with pleural effusion and splenic infarct (Figures 3 and 4).

During the 5th day of hospital stay she developed acute deep venous thrombosis (DVT) of the left lower limb (Figure 5). Doppler venous study confirmed thrombus in the left popliteal, femoral and external iliac veins. On further evaluation, ANA, Anti-dsDNA (1:80 dilutions) and Antiphospholipid antibody were had diffuse abdominal tenderness.

Fig. 1: Ulcers in the angle of mouth and tongue

Fig. 2: CT brain showing diffuse atrophic changes

Fig. 3: Left lower lobe atelectasis seen on chest X-ray

Fig. 4: CT chest showing left lower lobe atelectasis (arrow)
positive. A diagnosis of Systemic lupus erythematosus (SLE) with secondary antiphospholipid antibody syndrome (APLA) was made.

Since the patient complained of diminished vision, she was sent for ophthalmologic evaluation which showed Kayser–Fleischer (KF) ring on slit-lamp examination (Figure 6).

So, the patient was evaluated further for Wilson’s disease. Serum ceruloplasmin was found to be low i.e. 8 mg/dl (Normal 18 – 35 mg/dl) and a 24-hour urinary copper excretion was 1650 µg, hence Wilson’s disease was confirmed. So we concluded that it is a case of SLE with Wilson’s disease.

Discussion

Wilson’s disease, also known as hepatolenticular degeneration is an autosomal recessive disorder caused by mutation in ATP7B gene. The clinical presentation may be varied and it can involve multiple organs. It can present as unexplained neurological, psychiatric or liver problems. In fact psychiatric problem can be the earliest manifestation. More than 90 % of patients with neurological manifestations will have a KF ring.

Systemic lupus erythematosus is also a multisystem disorder and it can present with manifestations in any organ system, like skin and mucous membrane, heart, lungs, kidney, musculoskeletal, neuropsychiatric or haematological problems. People with SLE have an association with antiphospholipid antibody syndrome, which can lead to thrombotic manifestations. Neuropsychiatric manifestations in SLE is one of the challenging aspects in medicine. American college of Rheumatology has described 19 neuropsychiatric syndromes in SLE. Some of the common manifestations are headache, mood disorder, seizure and psychosis.

Our patient initially had chronic fever, following which she developed neuropsychiatric manifestations like altered behaviour and seizures. These symptoms are common to both SLE and Wilson’s disease. She had classical features of SLE like oral ulcers, lung collapse, normocytic anaemia, thrombosis of left lower limb veins, repeated abortions with ANA and dsDNA positive, with an associated APLA syndrome.

SLE in combination with Wilson’s disease is a rare presentation. No relation between these two conditions has been established. Most cases of SLE occur in Wilson’s disease patient secondary to D penicillamine therapy, in the form of drug-induced lupus.

We report this case for the varied clinical manifestations caused by two rare diseases occurring together in the same patient. Probably this case is the second of its kind, one case being reported from Korea which presented with SLE and acute liver failure in a 12 year old girl.

References