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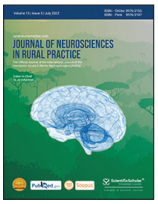

Letter to Editor

Altered behavior with an altered cell line

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Dear Editor,

Wilson's disease is an autosomal recessive disorder with a defect in ATP7B gene on chromosome 13q, causing defective copper incorporation and hence copper excess resulting in liver, neurological, eye, and other organ manifestations. Wilson's disease has been identified in both children and adults and has variable presentations. The clinical presentation may be as acute or chronic liver disease, progressive neurological disorder without clinically prominent hepatic dysfunction, isolated acute hemolysis, or psychiatric illness.[1,2] Wilson disease is found worldwide, with an estimated prevalence of 1 case/30,000 live births in most populations.[3] It is a rare condition; therefore, the diagnosis can be a challenge to most physicians as it can present with a multitude of symptoms affecting more than one system. A 35-year-old male, working in the IT sector, known case of hypothyroidism and social anxiety disorder, who presented with on and off bilateral pedal edema for 3 years. He has a history of alternative medicine intake, intermittently for 2 years. The patient had no history of drug or alcohol addictions. He had stable vitals on presentation, with bilateral pitting pedal edema, no pallor, icterus, cyanosis, clubbing, or lymphadenopathy. Cardiovascular examination showed loud second heart sound in pulmonary area. Other systems were within normal limits. Routine laboratories showed hemoglobin of 7.9 g/dL, macrocytic blood picture, platelets count of 0.45/mL, and total white cell count of 5460 cells/mm^3. Renal function test was within normal limit, and urine examination showed no proteinuria. Aspartate transaminase and alanine transaminase were 58U/L and 45U/L, respectively. Alkaline phosphatase - 45IU/L, serum albumin - 2.8 g/dL, serum globulin - 2.0 g/Dl, total protein - 4 g/dL, and mild indirect hyperbilirubinemia present with total bilirubin - 3.93 mg/dL. Peripheral smear showed normocytic normochromic anemia and thrombocytopenia with <0.5% schistocytes. The patient had in-hospital fever spikes for which fever panel was sent and dengue virus serology immunoglobulin M was positive. The patient had a non-fluid responsive hypotension and was shifted to intensive care unit and initiated on inotropic support. Serum cortisol was 1.78 mcg/dL, steroid replacement was given, and blood pressure improved. Initial ultrasound showed splenomegaly. An autoimmune hemolytic anemia was considered and antinuclear antibody (ANA), line immuno assay (LIA), antiphospholipid antibody (APLA), cytoplasmic antineutrophil cytoplasmic autoantibody (CANCA), and peripheral Antineutrophil cytoplasmic autoantibody (PANCA) were all negative. 2D echo done in view of the loud P2 showed dilated right atrium (RA), right ventricle (RV), ejection fraction (EF) - 58% and moderate pulmonary artery hypertension (PAH) -58 mmHg. Computed tomography (CT) thorax showed normal lung parenchyma, features suggestive of pulmonary hypertension and provisional diagnosis of idiopathic pulmonary hypertension was considered. Patient developed one-episode GTCS, following which the patient had cardiac arrest and was initiated on mechanical ventilator support. Magnetic resonance imaging of brain showed pituitary microadenoma with hemorrhage. Peripheral smear showed persistent schistocytes, increased lactate dehydrogenase, thrombocytopenia, low haptoglobin with a normal prothrombin time/activated partial thromboplastin time/international normalized ratio, a probable microangiopathic hemolytic anemia (MAHA) secondary to thrombocytopenic purpura was suspected. ADAMTS13 was low (37). Had 6 cycles of plasmapheresis done but thrombocytopenia persisted. Considering the low fibrinogen, elevated D-Dimer, and increased fibrin degradation products (FDPs), which persisted, a diagnosis of chronic disseminated intravascular coagulation (DIC) was made. Positron emission tomography–CT done showed no evidence of malignancy. Due to persistent abdominal pain, CT scan done showed paralytic ileus, splenomegaly, and mild ascites with chronic parenchymal liver disease. Hepatitis B and C serology and human immunodeficiency

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virus antibody were negative. Ascitic fluid analysis showed high serum ascitic-albumin gradient with white blood cell 37 leukocytes/microliter. Portal vein Doppler was within normal limits. Considering the presence of liver failure, with hemolysis and a background of psychiatric illness, with new-onset GTCS, a diagnosis of Wilson's disease was suspected. 24-h urine copper was 232 μg/24 h (elevated). Serum ceruloplasmin was 0.2 g/L (0.2–0.6). Slit lamp examination for Kayser–Fleischer ring was negative. Empirical chelation with D-penicillamine, zinc acetate was started as soon as the diagnosis was suspected. Transjugular liver biopsy showed features suggestive of chronic hepatitis, focal moderate intracellular pigment deposition seen, but no increase in dry weight of copper. The patient had a prolonged intensive care unit stay with multiple episodes of hepatic decompensation and was subsequently discharged after 3 weeks of hospital stay. On follow-up, the patient showed clinical improvement and showed reduction in 24-h copper excretion.

An autoimmune hemolytic anemia with thrombocytopenia due to severe dengue fever was considered as the initial differential. Autoimmune etiology was ruled out. At presentation, the patient had hemolytic anemia and hepatic failure and during the course of stay, neurologic symptoms ensued. The next differential in mind was neuro–Wilson's presenting as acute on chronic liver failure with porto pulmonary hypertension, chronic hepatic encephalopathy precipitated by alternative medicine or bile acid synthetase defect. The patient had a Leipzig score of 5 which was diagnostic of Wilsons. Serum ceruloplasmin was low normal, and 24-h urine copper was highly elevated. The background psychiatric disorder with the new-onset development of acute liver failure, hemolysis, and seizures was the diagnostic clues toward Wilson's disease. The challenge faced in this study was the atypical presentation of the disease. The liver biopsy was done after chelation was started as the patient needed to be stabilized before the procedure thus possibly interfering with the biopsy report. The main learning point was the atypical features of Wilson's disease and the various complications. Through this study, we would like to demonstrate the need to suspect Wilsons in young adults with psychiatric problems presenting with atypical hematological and liver manifestations.

Declaration of patient consent

Patient's consent is not required as the patient's identity is not disclosed or compromised.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The author(s) confirms that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Hedera P. Wilson's disease: A master of disguise. Parkinsonism Relat Disord 2019;59:140-5.
2. Nagral A, Sarma MS, Matthai J, Kukkle PL, Devarbhavi H, Sinha S, *et al*. Wilson's disease: Clinical practice guidelines of the Indian National Association for Study of the Liver, the Indian Society of Pediatric Gastroenterology, Hepatology and Nutrition, and the Movement Disorders Society of India. J Clin Exp Hepatol 2019;9:74-98.
3. Sandahl TD, Laursen TL, Munk DE, Vilstrup H, Weiss KH, Ott P. The prevalence of Wilson's disease: An update. Hepatology 2020;71:722-32.

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