Case Report

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Wilson's Disease Presenting as Status Epilepticus and Acute Fulminant Hepatic Failure in a Young Adult: A Case Report

Internal Medicine Section

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ABSTRACT

Wilson's disease (hepatolenticular degeneration) is a rare autosomal recessive disorder of copper metabolism caused by mutations in the ATP7B gene, resulting in pathological copper accumulation in the liver, brain, and other organs. Neuropsychiatric and hepatic manifestations may coexist or present in isolation, and the disease is frequently underdiagnosed due to its protean clinical features. A 19-year-old male presented with a one-week history of low-grade fever, frontal headache, and abdominal pain, complicated by Status Epilepticus (SE) with refractory generalised tonic-clonic seizures and a Glasgow Coma Scale (GCS) score of E1V1M2 on admission. Investigations revealed Acute Fulminant Hepatic Failure (AFHF) with markedly elevated transaminases {Serum Glutamate Pyruvate Transaminase (SGPT): 3938 U/L, Serum Glutamate Oxaloacetate Transaminase (SGOT): 3804 U/L}, coagulopathy (INR: 2.01), and hyperammonaemia (187.2 µg/dL). Magnetic Resonance Imaging (MRI) brain on epilepsy protocol demonstrated cortical and subcortical white matter hyperintensities consistent with metabolic encephalopathy. Diagnosis of Wilson's disease was established by a markedly elevated 24-hour urinary copper of 1069 mcg/24 hours (reference: 15-60 mcg/24 hours) and a subnormal serum ceruloplasmin of 18 mg/dL (reference: 20-60 mg/dL). The patient was managed with mechanical ventilation, multiple antiepileptic agents, empirical antimicrobials, N-acetylcysteine, and copper-lowering therapy with zinc sulfate and D-penicillamine. He was successfully extubated and discharged neurologically intact after eleven days of intensive care. At two-month follow-up, he remained seizure-free with significant clinical and biochemical improvement. This case highlights Wilson's disease as a rare but treatable cause of SE and acute liver failure in young patients. Early consideration of Wilson's disease in young adults presenting with combined neurological and hepatic dysfunction is critical, as prompt initiation of copper-lowering therapy can result in complete neurological recovery and prevent mortality.

Keywords: Ceruloplasmin, Copper metabolism, D-penicillamine

CASE REPORT

A 19-year-old male, employed as a plumber with no significant past medical or family history and no history of substance use, presented to the emergency department with a history of insidious onset low-grade fever associated with frontal headache and mild generalised dull aching abdominal pain for one week unresponsive to medication administered at a local health facility. The presenting complaint was further complicated by multiple episodes of generalised tonic-clonic seizures characterised by tonic-clonic movements of all four limbs, upward rolling of eyeballs, frothing at the mouth, and urinary incontinence since morning. These episodes recurred with an inter-ictal interval of approximately 1.5 hours without any recovery of consciousness between episodes, consistent with SE. Two episodes of vomiting containing food particles were also reported.

On presentation, the patient's GCS score was E1V1M2, indicating a deeply unconscious state. Vital signs were as follows: temperature 101.1°F, pulse rate 150 beats per minute (tachycardia), blood pressure 140/80 mmHg, and respiratory rate 20 breaths per minute (tachypnoea) with oxygen saturation of 80% on room air, indicating significant hypoxia. Neurological examination demonstrated normal tone in all four limbs, symmetrical deep tendon reflexes of grade +2 bilaterally in both upper and lower limbs, and bilaterally mute plantar responses. No meningeal signs were elicited. Ophthalmologic evaluation by slit-lamp examination revealed no papilledema, hypertensive retinopathy, or Kayser-Fleischer rings.

Laboratory investigations demonstrated a leukocytosis of 18,000 cells/cumm with normal haemoglobin (15.7 g/dL) and platelet count (3.09 lac cells/cumm). Liver function tests revealed markedly elevated transaminases (SGPT: 3938 U/L, SGOT: 3804 U/L) with a serum bilirubin of 3.7 mg/dL (direct 2.4 mg/dL, indirect 1.3 mg/dL), suggesting acute hepatocellular injury. Coagulation parameters were deranged with PT/INR of 28.2/2.01 and APTT of 37 seconds. Serum ammonia was elevated at 187.2 µg/dL (reference range: 20-122 µg/dL) [Table/Fig-1]. Ultrasonography of the abdomen and

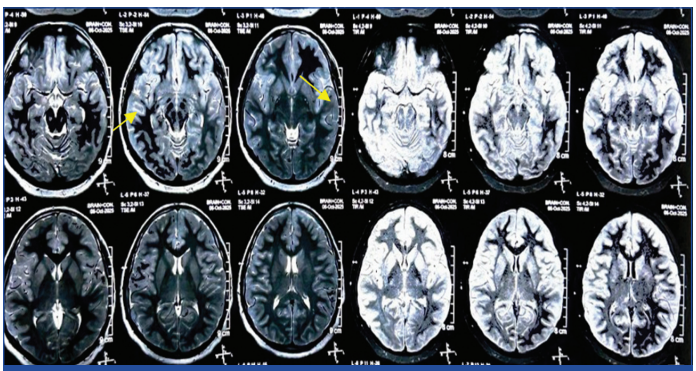
Reports	Day 1	2	3	4	5	6	7	8	9	10	11
Haemoglobin (g/dL)	15.7	15.8	15.8	14.8	14.9	15.4	15.5	15	14.3	14.9	14.8
Complete blood count (/cumm)	18000	11000	10000	11500	10300	9000	9800	8000	9000	7800	6100
Platelet count (lac/cumm)	3.09	2.75	2.75	2.4	2.42	2.5	2.6	2.7	2.5	2.68	2.7
Prothrombin time /INR sec	28.2/2.01	21/1.5	28.9/2.01	21/1.5	20/1.42	25	21.5/1.6			15/1.02	
Activated partial thromboplastin time sec	37	30	37.7	40	34	37.8	44			44	
Creatinine (mg/dL)	1	0.8	0.8	0.9	1.2	1.0	1.3	1	0.9	1	
Serum urea (mg/dL)	42	23	22	44	37	24	25	22	22	20	

S. bilirubin (mg/dL)	3.7 (2.4/1.3)	5.2 (3.4/1.8	5.5 (3.4/2.1)	3.6 (1.6/2)	2.9 (1.5/1.4)	2.6 (1.5/1.1)	2.8 (1.4/1.4)	2.5 (1.3/1.2)	1.5 (0.8/0.7)	1.6 (0.8/0.8)	
SGOT (IU/L)	3804	2500	770	294	126	194	172	170	164	60	
SGPT (IU/L)	3938	3300	2383	1533	907	501	410	302	360	117	
U/L		895								400	
S. uric acid (mg/dL)		7.8									
Serum sodium (meq/L)	134	140	142	146	138	139	142			148	
Serum potassium (meq/L)	3.6	4	4.5	3.4	4	5	4.4			3.9	
C-reactive protein (mg/L)	25.9		30		12.6					14	
Serum ammonia (20-122 µg/dL)	187.2		140				100			80	
Serum calcium (8.5-10.5 mg/dL)	8.2						8.5				
Serum magnesium (1.7-2.2 mg/dL)	3.2						2				
Serum iron (60-170 µg/dL)		88									
Serum ferritin (12-300 ng/mL)		297									
Total iron binding capacity (250-370 µg/dL)		344									
Serum triglyceride (<150 mg/dL)		241									
ESR		40									
24-hour urinary copper (15-60 mcg/24 hours)			1069								
Serum ceruloplasmin (range: 20-60 mg/dL),			18								

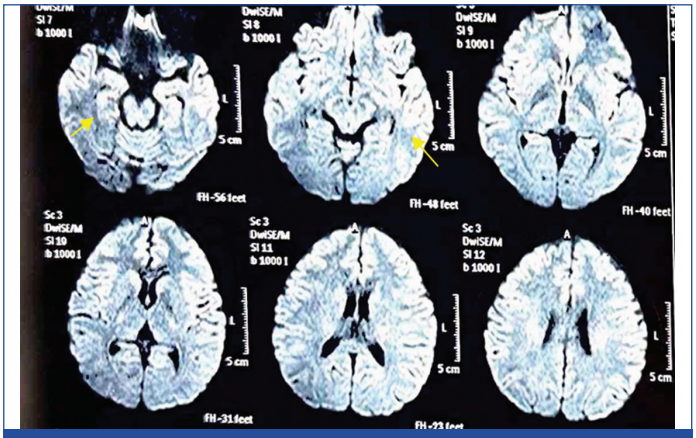
[Table/Fig-1]: Laboratory investigations during hospital stay.

pelvis suggestive of hepatomegaly with coarsened echotexture with splenomegaly with bilateral raised kidney echogencity and mild ascites. Infective workup including malaria, typhoid, dengue, hepatitis A, hepatitis E, and Leptospira IgM serology were all negative. Antinuclear Antibody (ANA) profile was negative, effectively excluding autoimmune hepatitis.

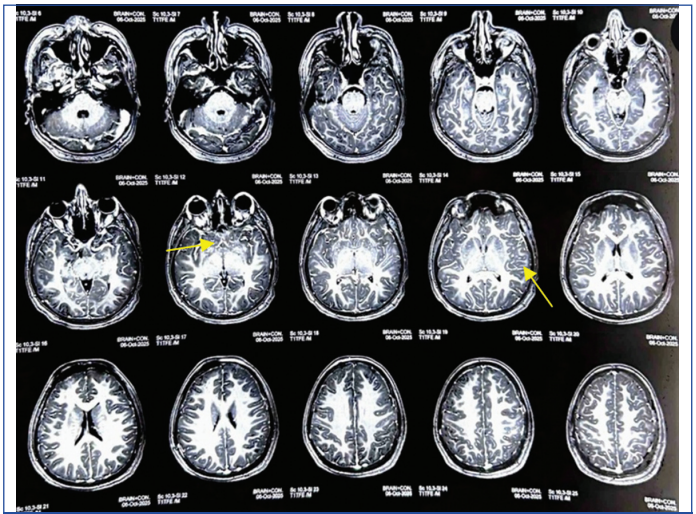
Cerebrospinal Fluid (CSF) analysis demonstrated a total cell count of two cells (100% lymphocytes), protein of 42 mg/dL, glucose of 59 mg/dL against a matched serum glucose of 90 mg/dL, and Adenosine Deaminase (ADA) of 0.4 IU/L. CSF Acid Fast Bacilli (AFB) smear and Cartridge Based Nucleic Acid Amplification Test (CBNAAT) were negative, ruling out neurotuberculosis. MRI of the brain performed on epilepsy protocol revealed multiple cortical and subcortical white matter hyperintensities on T2-weighted and Fluid-Attenuated Inversion Recovery (FLAIR) sequences, with restricted diffusion on Diffusion-Weighted Imaging (DWI), consistent with metabolic or toxic encephalopathy rather than an acute ischemic or inflammatory [Table/Fig-2-4]. The critical diagnostic investigations were the 24-hour urinary copper of 1069 mcg/24 hours (laboratory reference range: 15-60 mcg/24 hours), representing an approximately 18-



[Table/Fig-2]: Axial MRI brain: a) T2W and b) FLAIR sequences showing cortical and subcortical white matter hyperintensities bilaterally, consistent with metabolic or toxic encephalopathy.



[Table/Fig-3]: Axial DWI MRI brain showing restricted diffusion in the cortical and subcortical white matter bilaterally.



[Table/Fig-4]: Axial T1-weighted MRI brain showing hypointense signal in the cortical and subcortical white matter bilaterally.

fold elevation, and serum ceruloplasmin of 18 mg/dL (laboratory reference range: 20-60 mg/dL), which was below the lower limit of normal. These findings in conjunction with the clinical presentation established the diagnosis of Wilson's disease.

Given the initial presentation, the patient was intubated and mechanical ventilation was commenced in view of hypoxia, reduced GCS, and refractory seizures. Midazolam infusion (20 mcg/kg/hour) was started for seizure control at least for 24 hours which was gradually tapered off and switched to three antiepileptic agents: intravenous levetiracetam 1 gram twice daily, intravenous lacosamide 100 mg twice daily, and tablet clobazam 10 mg twice daily. Broad-spectrum empirical antimicrobial coverage was instituted with intravenous ceftriaxone two grams twice daily, vancomycin 1 gram twice daily (15 mg/kg/day), and intravenous acyclovir 1 gram thrice daily (15 mg/kg/8 hours) in view of the possibility of bacterial meningitis or herpes simplex encephalitis, in accordance with the Infectious Diseases Society of America (IDSA) clinical practice guidelines for the management of encephalitis [1]. Oral doxycycline 100 mg twice daily was added considering atypical organisms as a cause for persistent fever. Management of acute fulminant liver failure included intravenous N-acetylcysteine infusion over 24 hours, syrup lactulose, and lactulose enema to promote intestinal clearance of ammonia.

Following confirmation of Wilson's disease, copper-lowering therapy was initiated with tablet zinc sulfate 50 mg thrice daily. D-penicillamine was subsequently planned and initiated 250 mg four times a day along with pyridoxine 25 mg/day. The patient was gradually weaned from mechanical ventilation and successfully extubated. He regained full consciousness and orientation progressively, becoming ambulatory with minimal support. Following eleven days of critical care admission, the patient was shifted to ward. At the time of discharge, the patient was haemodynamically stable with a GCS of 15/15 (E4V5M6), afebrile, maintaining adequate oxygen saturation on room air, and ambulatory with minimal support. Repeat liver function tests from fourth day till discharge showed a downward trend in transaminases, reflecting hepatocellular recovery in response to copper-lowering therapy, though values remained above normal limits. Coagulation parameters showed partial correction with INR improving from 2.01 at admission to near-normal value of 1.02 on discharge. Serum ammonia levels had normalised. The patient was tolerating oral feeds well with no nausea or vomiting, and bowel habits had normalised. The patient was advised to continue zinc supplementation and D-penicillamine along with pyridoxine vitamin b complex on a long-term basis.

At the two-month follow-up visit, the patient presented in a markedly improved general condition. He was fully oriented to time, place, and person, conversing fluently, and had resumed light activities at home. He reported no recurrence of seizures throughout the follow-up period. There was no complaint of tremors, dystonia, or any new neurological symptoms. Gastrointestinal tolerance to D-penicillamine had improved significantly, and the patient reported no adverse effects. He remained compliant with dietary copper restriction and the prescribed medication regimen.

DISCUSSION

Wilson's disease is an autosomal recessive disorder caused by pathogenic variants in the ATP7B gene, resulting in defective biliary copper excretion and impaired ceruloplasmin synthesis, leading to progressive copper accumulation predominantly in the liver, brain, and other organs. The clinical presentation is heterogeneous, encompassing hepatic, neurological, psychiatric, or mixed manifestations [2]. Neurological involvement is characteristically extrapyramidal, including tremor, dystonia, dysarthria, and Parkinsonism, while epileptic seizures remain relatively uncommon. When present, seizures are typically generalised tonic-clonic, though focal seizures and SE have been described. Proposed mechanisms include direct neurotoxicity of free copper, metabolic disturbances from hepatic dysfunction, and treatment-related pyridoxine deficiency [2-4]. Early psychiatric manifestations such as psychosis, personality changes, and cognitive decline may precede or overshadow hepatic dysfunction, significantly delaying diagnosis [5,6].

The simultaneous presentation of SE and AFHF as the index manifestation of WD, as observed in the present case, is exceptionally rare. AFHF in WD is characterised by markedly elevated transaminases, hyperbilirubinaemia, coagulopathy, and hyperammonaemia, frequently accompanied by Coombs-negative haemolytic anaemia, and may rapidly progress to multi-organ dysfunction [2]. The co-existence of AFHF and SE creates a significant diagnostic challenge, as hepatic encephalopathy, infectious meningoencephalitis, and autoimmune encephalitis must be systematically excluded. In the present case, broad-spectrum empirical antimicrobial and antiviral coverage was instituted with intravenous ceftriaxone, vancomycin, and acyclovir in accordance with IDSA encephalitis management guidelines [1], while infectious and autoimmune aetiologies were excluded by non-inflammatory CSF analysis and negative serological workup. Multifocal cortical and subcortical MRI hyperintensities on T2 and FLAIR sequences, without restricted diffusion, were consistent with metabolic encephalopathy rather than an ischaemic or inflammatory aetiology, which together with markedly elevated 24-hour urinary copper and low serum ceruloplasmin, ultimately established the diagnosis of WD.

Diagnosis of WD requires integration of clinical, biochemical, and radiological parameters. Serum ceruloplasmin below 10 mg/dL carries a high positive predictive value, while 24-hour urinary copper exceeding 100 µg/day in symptomatic patients remains one of the most sensitive screening parameters. Non-ceruloplasmin-bound free copper serves a useful adjunct role, primarily in treatment monitoring. Kayser-Fleischer rings detected on slit-lamp examination are a critical clinical clue, particularly in neurological WD. MRI brain is superior to CT, with characteristic T2 signal alterations in the basal ganglia, thalamus, and midbrain; the pathognomonic "face of the giant panda" sign, though infrequently observed, is highly specific. A normal MRI effectively excludes neurological WD [7,8]. In the present case, the absence of Kayser-Fleischer rings and the predominance of AFHF with SE initially obscured the diagnosis, highlighting that WD can manifest without classic features, necessitating a high index of suspicion in young patients with unexplained acute liver failure and neuropsychiatric manifestations.

Management of WD centres on reducing copper accumulation through chelation or intestinal copper blockade. D-penicillamine at 20 mg/kg/day promotes urinary copper excretion and is conventionally co-administered with pyridoxine 25 mg/day given its weak anti-pyridoxine activity. Trientine serves as an alternative for penicillamine-intolerant patients, while zinc salts induce intestinal metallothionein to prevent systemic copper absorption, suited particularly for asymptomatic patients or maintenance therapy. Combination zinc and D-penicillamine therapy, as employed in the present case, has demonstrated efficacy even in severe presentations. Lifelong maintenance therapy is mandatory to prevent rebound copper accumulation [7,8]. Despite critical illness requiring mechanical ventilation, the present patient achieved dramatic neurological recovery following initiation of targeted anti-copper therapy, reinforcing that even severely ill patients with WD-related AFHF can attain favourable outcomes with timely intervention.

Several published case reports describe comparable but distinct presentations. Leelamani V et al., described a young male with acute encephalopathy and SE initially treated as encephalitis before WD was confirmed on follow-up evaluation [4], mirroring the diagnostic delay observed in the present case. D'Andrea L et al., reported a functional seizure presentation in WD [5], while Buciuc

AG et al., described WD masquerading as psychosis and catatonia with concealed hepatic dysfunction [6], both highlighting the broad neuropsychiatric spectrum of WD. Elendu C et al., highlighted ATP7B-confirmed WD with neuropsychiatric predominance where chelation reversed antipsychotic-refractory symptoms [9]. Kumar S reported a 16-year-old girl with SE, low ceruloplasmin, and elevated urinary copper achieving rapid recovery with D-penicillamine [10], most closely paralleling the present case in terms of SE as the presenting feature. Rasib AR et al., reported recurrent seizures with cerebellar dysfunction previously misdiagnosed as encephalitis, with complete seizure control achieved after anti-copper therapy [11]. Collectively, these cases corroborate that WD presenting with seizures and encephalopathy is frequently misdiagnosed, and that early copper-specific therapy is pivotal for neurological recovery. The present case is distinctive in representing a rare and critical confluence of SE, AFHF, respiratory failure, and severe coagulopathy as the inaugural presentation of WD in a young adult, with complete neurological recovery underscoring the lifesaving potential of timely diagnosis and targeted management.

CONCLUSION(S)

This case highlights Wilson's disease as a rare but important cause of acute fulminant liver failure presenting with SE in a young adult. The nonspecific initial symptoms and overlapping features with hepatic encephalopathy and meningoencephalitis posed a significant diagnostic challenge. A high index of suspicion and timely evaluation of copper metabolism were crucial in establishing the diagnosis after common infectious and autoimmune causes were excluded. Early initiation of anti-copper therapy resulted in dramatic neurological and clinical recovery, avoiding further deterioration. This report emphasises the need to consider Wilson's disease in young patients with unexplained acute liver failure and neuropsychiatric manifestations. Prompt recognition and targeted treatment can be lifesaving and lead to favourable outcomes.

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