

**Missing Wilson's is Easy and Deadly!!**G Vishwa Reddy<sup>1\*</sup>, Ashima Shama<sup>2</sup>, Raghu Kondle<sup>3</sup><sup>1</sup>Assistant Professor, Department of Emergency Medicine, Nizam's Institute of Medical Sciences, Hyderabad - 500082, Telangana State, India<sup>2</sup>Professor & HOD, Department of Emergency & Critical Care Medicine, Nizam's Institute of Medical Sciences, Hyderabad - 500082, Telangana State, India<sup>3</sup>Head of Clinical Services & Senior Consultant, Department of Emergency & Critical Care Medicine, Narayana Medical College and Hospital, Nellore, Andhrapradesh, India.**\*Corresponding author**

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**Article History**

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**Abstract:** We report a case of a young male child with chief complaints of altered sensorium and involuntary movements of the limbs presented to emergency and diagnosed with Wilson's disease. Complaints of neurological, physiological and biochemical parameters were supported to easy diagnosis to setup the management/treatment modalities.**Keywords:** Wilson's disease, copper excretion, neurological parameters.**INTRODUCTION**

Wilson's disease is an autosomal recessive disorder caused by mutation in ATP 7B gene. The disease results in impaired copper excretion by liver. Finally copper will deposit in the liver, brain, which causes tissue damage. Wilson's disease is a disease of adolescence, 50% presenting with hepatic disease as the first disease manifestation [1].

**CASE REPORT**

A 13 year old male presented to ED with (chief) complaint(s): c/c altered sensorium since two days, involuntary movements of the limbs.

**Primary survey**

Airway – Patient:

Breathing- RR-26/min; Spo2- 96 %- WNL

Circulation- pulse- 120/min; B.P.- 130/90-WNL.

Disability- patient in active seizure episode with tonic posturing of the limbs

**Interventions done**

Airway secured.

Injected midazolam 2mg given for control of seizure episode, followed by injected leviteracetam -500mg.

**Focused history of present illness**

Patient was apparently asymptomatic one day before, followed by altered behavior in the form of irritability; not obeying commands, involuntary movements of the limbs; tonic posturing of the limbs and associated up rolling of eye balls;

There was no h/o of fever; vomiting; headache; bleeding manifestations; cough; No h/o of decreased urine output; burning micturition; shortness of breath and chest pain.

**Past history**

No h/o of similar complaints in the past.

No h/o of epilepsy, hypertension; diabetes; tuberculosis and asthma.

No h/o of CVA; CAD; CKD

**Personal history**

Mixed diet, decreased appetite, non-alcoholic and non-smoker, normal bowel and bladder habits

**Family history**

No relevant family h/o

**Differential diagnosis based on history**

- Status epilepticus
- Meningitis
- Encephalopathy secondary to metabolic disorder / sepsis

**Examination****Vital signs**

Pulse- 120/min; B.P – 130/90mmHg; RR-26/min; Spo2- 96 %- Temp-98.6F.GRBS- 126mg/dl

**General physical exam**

O/E Patient in status epilepticus, Icterus present, (+) No E/O of pallor; cyanosis; clubbing; Lymphadenopathy, kayser fleischer rings –present around the cornea.

**Focused systemic exam**

CNS- Patient in active seizure episode. with uprolling of eye balls and tonic posturing of the limbs, not responding to pain, increased tone in all the limbs, reflexes- present; plantar – extensor response. There was no neurological deficit  
 CVS– S1S2 present; No murmur  
 RS – bilateral air entry present, no adventitious sounds.

GIT – P/A Soft, distended, gross splenomegaly present, no hepatomegaly

**Provisional diagnosis/ differentials**

- Status epilepticus
- Viral meningitis with chronic liver disease
- Encephalopathy secondary to metabolic disorder / sepsis
- Wilsons disease

**Relevant investigations/adjuncts**

CT Brain - WNL, EEG- WNL  
 MRI Brain- hyperintense lesions were present in basal ganglia  
 HBsAg, anti-HBc IgM, anti-HAV IgM and anti-HCV antibodies- Negative

**Table-1: Biochemical and pathological diagnosis of the patient**

Parameter	Report
Serum copper	300 µg /dl
Urinary Copper	1200 µg/dl
Serum celluloplasmin levels	8 µg/dl
PT	14
INR	1.6
APTT	32
Blood urea	22 mg/dl
Serum creatinine	0.9 mg/dl
Serum electrolytes	
Na	130 mg/dl
k+	3.2mg/dl
cl-	100 mg/dl
Ca2+	9.3mg/dl, 3.2
phosphorous	mg/dl
Mg2+-	2 mg/dl.
ABG	
Ph	7.46
pCO2	28.2
Po2	127
HCO	20
CBP	
Hb	12gm/dl, 6200,
tlc	1,20000/cumm
platelet count	
LFT	
Total bilirubin	3.3 mg/dl
direct bilirubin	1.1 mg/dl
Indirect bilirubin	2.2 mg/dl
AST	96 U/L
ALT	73U/L
Alkaline phosphatase	141 U/L
Albumin	4.2 gm/dl
Globulin	3.3gm/dl

USG abdomen– Cirrhosis of liver, grade- 1 oesophageal varices

Ultrasound-assisted percutaneous liver biopsy report showed liver copper content was elevated - 55.68 µg/g dry weight.

**Final diagnosis**

Status dystonicus with chronic liver disease with portal hypertension secondary to Wilson's disease

**Clinical pearls**

Missing WILSONS in E.D is easy and dangerous as both misdiagnosis and delay in treatment would lead to severe neurologic disability, hepatic failure and death. Wilson's disease should be suspected in any young patient who presents with, neurological complaints like status dystonicus/status epilepticus even if Kayser Fleischer rings are absent and serum ceruloplasmin levels are normal, especially if other alternative diagnoses have been ruled out [2].

**REFERENCES**

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