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Missing Wilson's is Easy and Deadly!!

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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 paramters.

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: Bichemical 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 k+ cl- Ca2+ phosphorous Mg2+-	 130 mg/dl 3.2mg/dl 100 mg/dl 9.3mg/dl , 3.2 mg/dl 2 mg/dl.
ABG Ph pC02 Po2 HCO	 7.46 28.2 127 20
CBP Hb tlc platelet count	 12gm/dl, 6200, 1,20000/cumm
LFT Total bilirubin direct bilirubin Indirect bilirubin AST ALT Alkaline phosphatase Albumin Globulin	 3.3 mg/dl 1.1 mg/dl 2.2 mg/dl 96 U/L 73U/L 141 U/L 4.2 gm/dl 3.3gm/dl

USG abdomen– Cirrhosis of liver, grade- 1 oesohageal 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 dystonicius/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].

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