Challenges in Recognizing and Diagnosing Wilson Disease, Case Report

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BACKGROUND

- Wilson disease is a rare autosomal recessive disease caused by mutation in ATP7B causing copper deposit in the liver, brain and other organs.
- It is manageable, and physicians must familiarize themselves with the diversity of its presentation.

CASE DESCRIPTION

- A 58 years old female presented with acute psychosis and altered mental status. Patient was admitted with suspected hepatic encephalopathy.
- She developed **Acute liver failure** with **Coombs negative hemolytic Anemia**
- She had history of liver cirrhosis, COPD, and Alpha-1 Antitrypsin deficiency.

PAST DISEASE COURSE

- She was diagnosed with liver cirrhosis in 2019 when presented with Ascites, her work up showed
  - **Ceruloplasmin of 13**
  - ALP 247, AST/ALT 51/16, total bilirubin 0.7
  - ANA >1: 1280
  - Anti smooth muscle Ab positive
  - Antimitochondrial Ab positive.
  - **Alpha 1 antitrypsin low 72 with MZ phenotype**
- Liver biopsy 2 years earlier consistent with cirrhosis NAS stage 4/4 showing moderately active steatohepatitis with NAS stage IV, no copper staining used
- Recurrent admission with **acute psychosis** and **slurred speech, tremor** and **choreiform like** movement

HOSPITAL COURSE

- She developed acute liver failure with **MELD score 36%**
- **Liver failure and Coombs negative hemolytic anemia** triggered the possibility **of Wilson disease** as the cause of her cirrhosis
- **Mental status** deteriorate and transferred to ICU for intubation
- She developed multiorgan failure and started on CRRT
- We were unable to perform slit lamp due to her condition
- MRI of brain showed intracranial hemorrhage.
- Developed Fungemia with increased on pressor requirements
- Eventually family decided on comfort care and patient passed away

Wilson Disease is a rare autosomal recessive condition. **Atypia of age** of presentation can cause challenges at recognizing it. **Leipzig scoring system** helped to identify high risk patient

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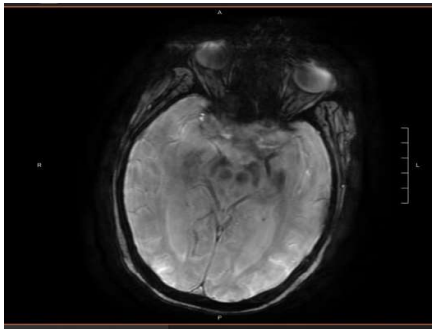
IMAGE MRI



Panda eye sign on MRI is a feature resulting from the sparing of the red nuclei within a diffuse midbrain hyperintensity(3).

CURRENT ADMISSION LABS

TEST	Patient Value	Reference range
Ceruloplasmin	5.9 mg/dl	19-39
24 hour urine copper (60 ml)	351 mcg	3-35
Copper serum	37	80-185
Haptoglobin	< 8 mg/dl	<8
Hemoglobin	6.7 mg/dl	12-16
ALT	59 unit/L	12-78
AST	171 unit/L	15-37
ALP	541	45-117

DISCUSSION

The diagnosis is based on the score developed at the 8th International Meeting on Wilson Disease in **Leipzig** (2). If the score is ≥4, Wilson disease is highly likely. In our patient, points are given as follows:

- Neuropsychiatric symptoms suggestive of Wilson disease (2 points),
- Coombs-negative hemolytic anemia (1 point),
- Serum ceruloplasmin < 10 mg/dL (2 points)
- **Total score of 5 points**, which made her diagnosis with Wilson disease is highly likely.

REFERENCE

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