

Question 1 – August 31

A 20 year old woman presents for evaluation of abnormal liver tests. She has exhibited bizarre behavior for several months and left college. She denies alcohol and substance abuse. Family history is negative for liver disease. Physical exam is significant for an enlarged liver left lobe 6 cm below the xyphoid and a palpable spleen tip. Wilson's disease is suspected.

Lab data:

AST 152 U/L, ALT 64 U/L, alkaline phosphatase 18 U/L, albumin 3.1 g/dl, total bilirubin 1.7 mg /dl, Hemoglobin 8.2, Platelets 128, INR 1.6, HCV Ab negative, HBV sAg negative

Which of the following will support the diagnosis of Wilson's disease:

- A. Low serum ceruloplasmin
- B. Corneal rings on slit lamp examination
- C. Low alkaline phosphatase
- D. Rhodanine positive granules in hepatocytes on liver biopsy
- E. Low 24 hour urinary copper excretion

Answer: E

Urinary copper excretion is typically above 100 micrograms. The other answers are typical findings in Wilson's disease.

References:

EASL Clinical Practice Guidelines: Wilson's disease. European Association for Study of Liver. J Hepatol. 2012 Mar;56(3):671-85.

AASLD Practice Guidelines: Wilson's Disease. Roberts E, et al. Hepatology 2008.

Diagnosis and phenotypic classification of Wilson disease. Ferenci P, et al. Liver Int. 2003 Jun;23(3):139-42.