**Question 36 – April 10**

Which of the following statements is true regarding Wilson's disease?

A. Elevated ceruloplasmin is suggestive of the diagnosis.
B. Urinary copper levels are decreased in this disease.
C. Liver biopsy showing elevated copper levels is diagnostic.
D. Liver transplantation is the only cure.
E. Zinc salts or D-Penicillamine will cure this disease.

**Answer: D**

Wilson's disease is due to copper overload from a lack of ATP7B within hepatocytes. The protein ATP7B is important in the vesicular pathway of hepatic copper transport into bile. Gene mutation leads to absence or diminished function of ATP7B, resulting in a decrease in biliary copper excretion and ultimately to hepatic accumulation of copper. There is also reduced incorporation of copper into ceruloplasmin. Therefore, the only cure for this is transplantation of a liver that has functioning ATP7B. Serum ceruloplasmin concentration has a normal range of 20 to 50 mg/dL.

Approximately 95% of homozygous Wilson's disease subjects have values <20 mg/dL. Of note, 5% of homozygous subjects have normal ceruloplasmin while 20% of asymptomatic WD heterozygotes will have low ceruloplasmin.

Urinary copper levels are elevated in Wilson's disease due to increased levels of free, unbound copper and usually exceed 100 μg/24 hr in Wilson's disease due to increased levels of free, unbound copper and usually exceed 100 μg/24 hr in Wilson's disease. Elevated copper levels on liver biopsy can also be seen in PSC or PBC but not to the same degree of elevation as would be seen in Wilson's disease.

Zinc salts and D-pencillamine only chelate and maintain copper within the body. Therapy must be continued lifelong or there will be a re-accumulation of copper over time.

**Reference:**

Gitlin JD. Wilson disease. Gastroenterology 2003;125:1868-1877.