Question 49 – Week of June 6

A 24 year old male is referred for abnormal liver enzymes. He takes no medications other than albuterol for asthma. He denies any alcohol, illicit drug use or sexual activity. There has been no recent travel. His physical exam shows no stigmata of chronic liver disease. There is no scleral icterus, hepatomegaly, or asterixis. Serologies for viral hepatitis and autoimmune disease are negative. Additional testing shows A1AT level 130, iron saturation 25%, ferritin 120, and ceruloplasmin 10.

Which of the following is the most appropriate next step?

A. Send him to the ophthalmologist for slit lamp examination.
B. Send for the genetic test for Wilson disease.
C. Send urine for 24 hour copper excretion
D. Perform liver biopsy

Answer: C

There is no single confirmatory test for Wilson disease. The diagnosis is made using a combination of clinical, histologic and biochemical data. It should be considered in any individual between the ages of 3 and 55 years with liver abnormalities of uncertain cause. The simplest screening tests are slit lamp examination and serum ceruloplasmin level. Most patients will have a low ceruloplasmin (<20 mg/dL). More than half of subjects with isolated hepatic manifestations without neurologic symptoms do not have K-F rings, so their absence does not exclude Wilson disease. Urinary copper excretion usually exceeds 100 μg/24 hours in symptomatic patients and is the next step in this patient's evaluation. Additional testing includes increase serum copper (> 80 μ/dl), and increase serum free copper (> 200 μg/dl).