Diagnosing Wilson Disease: Perseverance is a Virtue

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Background

- Wilson disease is an autosomal recessive disorder with hepatic, neurologic and psychiatric symptoms.
- Prevalence from 1: 30,000 to 1: 66,000
- Approximately 15% of patients present with isolated hepatic dysfunction.
- We present a case of a young male with decompensated cirrhosis found to have Wilson disease via cumulative copper staining on liver biopsy

Case Description

- 35-year-old male with decompensated cirrhosis and ascites. Previously diagnosed at OSH ~1 year ago presumed etiology - NASH given BMI 48 and unremarkable labs.
- AST 318, ALT 160, Alk Phos 180, T bilirubin 16, and MELD-Na 25.
- Normal ceruloplasmin level 27.4mg/dL.
- Given the presumed diagnosis of decompensated cirrhosis secondary to NASH, the elevated transaminases were unusual.
- 24-hour urine copper 137mcg/day was elevated (normal 15-60).
- Slit lamp exam negative for KF rings.
- Transjuglar liver biopsy with marked cholestasis, hepatocyte injury, mild to moderate portal/septal inflammation and stage 4 nodule formation.

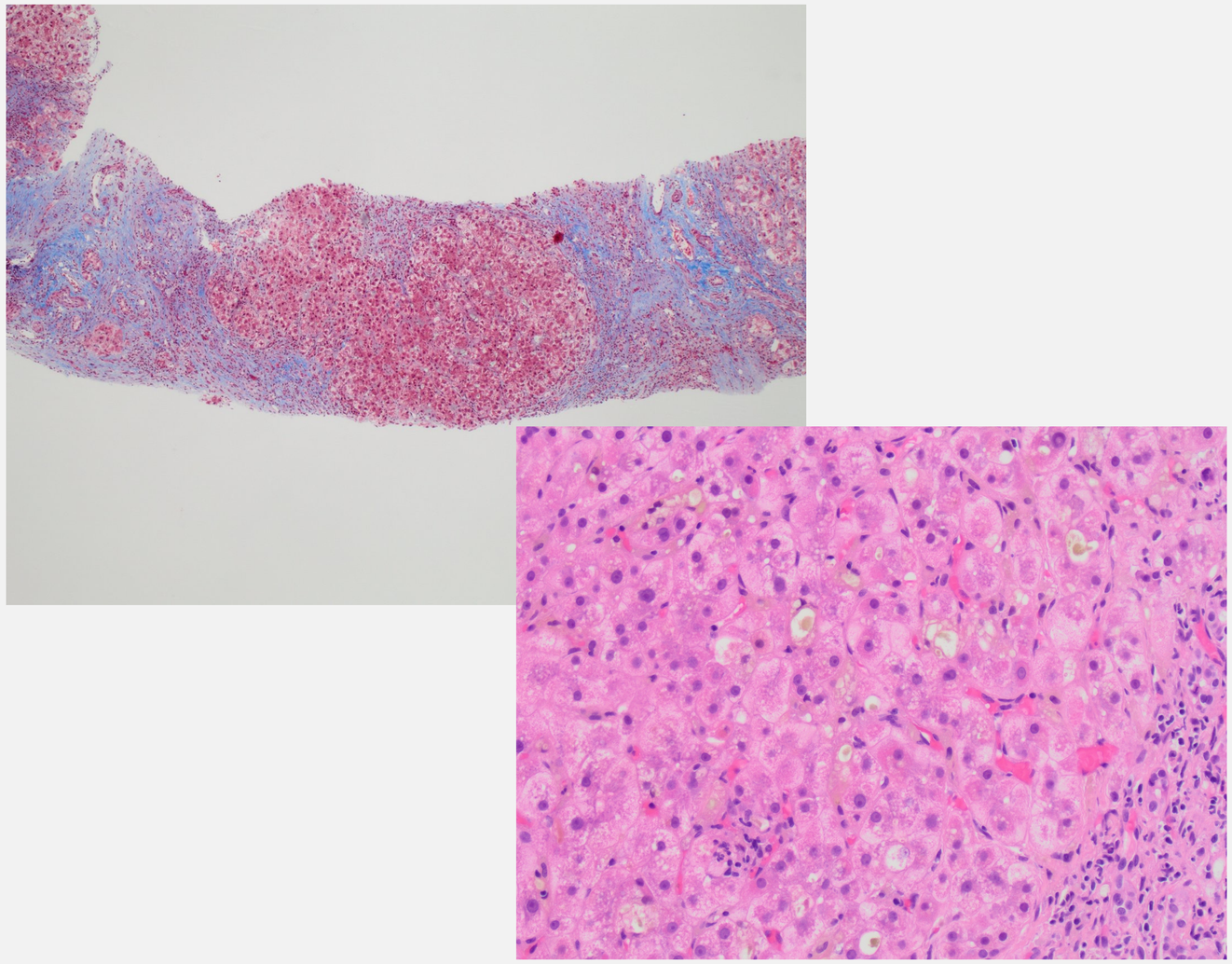

Figure: Sections show marked cholestasis with associated hepatocyte swelling. Large septal areas show bile ductular reaction with mixed septal inflammation. Trichrome stain highlights cirrhotic nodules (stage 4/4).

Case Description - continued

- A trial of prednisone 40mg daily and ursodiol 500mg TID started for possible autoimmune hepatitis.
- Cumulative copper stain on liver biopsy = elevated copper level 531mcg/g (normal <50).
- Prior to starting a copper chelating agent, patient was readmitted with MELD- Na 32 and underwent a successful liver transplant.
- He has since followed up in clinic without complications.

Discussion

- Wilson disease should be considered in adults with NASH< 35 years old, AST:ALT ratio>2, family history, or concomitate neurologic or psychiatric symptoms.
- Initial work up = serum ceruloplasmin levels, slit lamp exam, and 24-hour urine copper excretion.
- Liver biopsy or genetic testing when 24-hour urine copper is elevated without elevated ceruloplasmin levels or KF rings.
- KF rings are present in 90-99% with neurologic symptoms but 50% of patients with hepatic dysfunction.
- Our patient met Leipzig criteria with 24 hour urine copper >2 times ULN and total liver copper >5 times ULN; genetic mutation is unknown.
- Cumulative copper level on liver biopsy >250mcg/g is ~83% sensitive to diagnosis Wilson disease.
- Our case demonstrates the need for further evaluation in patients with newly diagnosed cirrhosis of uncertain etiology as Wilson Disease can be diagnosed without KF rings and normal ceruloplasmin levels.

References

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