Kayser-Fleischer rings of acute Wilson's disease

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Here we describe a case of a 22-year-old woman who presented with acute liver failure and Kayser-Fleischer rings suggesting the diagnosis of Wilson's disease.

A 22-year-old woman with no known past medical history presented to an emergency department with a 3-week history of fatigue, decreased appetite, and jaundice. Her initial laboratory workup indicated acute liver injury with a total bilirubin level of 12.1 mg/dL (reference range, 0.2–1.0); alkaline phosphatase of 45 U/L (reference range, 50–136); aspartate aminotransferase of 111 U/L (reference range, 15–37); alanine aminotransferase of 27 U/L (reference range, 12–78); ceruloplasmin of 19 mg/dL (reference range, 20–60); and prothrombin time of 29.11 seconds (reference range, 9.0–12.0).

Figure. Kayser-Fleischer ring in a patient with acute liver failure from Wilson's disease.

Additionally, she was found to have concurrent acute kidney injury and a Coombs-negative hemolytic anemia.

The patient underwent a laparoscopic cholecystectomy at an outside hospital that revealed the presence of ascites and a nodular liver with an otherwise normal gallbladder. The patient quickly decompensated into acute liver failure during her post-operative course, and physical examination revealed the presence of Kayser-Fleischer (KF) rings (*Figure*).

The presence of KF rings and jaundice with abnormal liver function tests, including low ceruloplasmin and alkaline phosphatase, suggested the diagnosis of Wilson's disease. The patient was transferred to Baylor University Medical Center at Dallas, where she underwent evaluation for orthotopic liver transplantation, which was successfully performed on hospital day 4. Elevated hepatic copper (2145 mcg/g; reference range, 10–35) on dry weight liver biopsy was consistent with the diagnosis of Wilson's disease. Mutation analysis indicated that the patient was homozygous for the pathogenic mutation 3201 C>A in exon 14, resulting in the amino acid change His1069Gln.

DISCUSSION

Wilson's disease is an autosomal recessive mutation of the *ATP7B* gene, whose protein both traffics excess copper into the bile canaliculus for excretion and binds copper to apoceruloplasmin, forming holoceruloplasmin. Oxidative damage ensues when uncomplexed copper begins to accumulate in hepatocytes. Liver injury can present in various forms ranging from asymptomatic abnormal liver function tests to acute and chronic liver failure. With ongoing hepatic damage, uncomplexed copper is released from the liver and deposited in other organs, including the lenticular nucleus of the basal ganglia, resulting in Parkinson-like symptoms; the proximal renal tubule, resulting in Fanconi's syndrome; and the red blood cells, resulting in a Coombs-negative hemolytic anemia. Other organs affected include the heart, pancreas,

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parathyroid gland, and bone. The eye is uniquely affected, as copper accumulates in Descemet's membrane of the corneal limbus, resulting in KF rings (*Figure*), and in the lens, resulting in sunflower cataracts. The observation of the characteristic brown-gold-yellow KF ring at the margin of the cornea and sclera generally requires a slit-lamp examination, but some rings can be seen under normal light. Each of these ocular findings resolves over time following transplant (1, 2).

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