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WILSON'S DISEASE

ABSTRACT

The Wilson's disease (WD) gene is located on chromosome 13. Deficiency of the WD gene product is likely to be responsible for the lack of copper incorporation into ceruloplasmin and the defective biliary excretion of copper in WD. The majority of symptomatic WD patients present with hepatic or neuropsychiatric features; the principal hepatic manifestations include fulminant hepatic failure, chronic hepatitis, and cirrhosis. In patients with a low serum ceruloplasmin, diagnosis of WD in the absence of Kayser-Fleischer rings requires determination of hepatic copper concentration. Serum detection of radiocopper incorporation into ceruloplasmin may be a useful alternative test when liver biopsy is contraindicated. The use of DNA marker studies is limited largely to genetic screening of young family members of difficult diagnostic situations, using the index patient's DNA as a reference. The drug of choice for treating WD patients is D-penicillamine, but alternatives under selected circumstances include zinc, or tetrathiomolybdate. Liver transplantation is indicated for patients with fulminant hepatitis or decompensated cirrhosis unresponsive to therapy.