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ABSTRACT

Wilson's disease is a genetic disorder in which copper builds up in the body and is an autosomal recessive condition due to mutation in the ATP7B . Symptoms are typically related to the brain & liver . Liver related symptoms include vomitings , weakness , fluid buildup in the abdomen , swelling of legs , yellowish skin & Itchiness . Brain related symptoms include tremors, muscle stiffness ,trouble speaking ,personality changes , anxiety. Diagnosis include liver function test , Magnetic Resonance Imaging (MRI), Liver biopsy , CT scan , Urinary copper excretion, serum copper , serum ceruloplasmin ,liver copper concentration . Treatment includes dietary changes , chelating agents, zinc supplements , liver transplantation

KEY WORDS: Mutation , ATP7B , chelating agents , serum ceruloplasmin.

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