Int Rev Neurobiol. 2013;110:313-48. doi: 10.1016/B978-0-12-410502-7.00014-4.

Update on Wilson disease.

Aggarwal A[1], Bhatt M.

Author information

Abstract

Wilson disease (WD) is an inherited disorder of chronic copper toxicosis characterized by excessive copper deposition in the body, primarily in the liver and the brain. It is a progressive disease and fatal if untreated. Excessive copper accumulation results from the inability of liver to excrete copper in bile. Copper is an essential trace metal and has a crucial role in many metabolic processes. Almost all of the body copper is protein bound. In WD, the slow but relentless copper accumulation overwhelms the copper chaperones (copper-binding proteins), resulting in high levels of free copper and copper-induced tissue injury. Liver is the central organ for copper metabolism, and copper is initially accumulated in the liver but over time spills to other tissues. WD has protean clinical manifestations mainly attributable to liver, brain, and osseomuscular impairment. Diagnosis of WD is challenging and based on combination of clinical features and laboratory tests. Identification of various high-frequency mutations identified in different population studies across the world has revived interest in developing DNA chips for rapid genetic diagnosis of WD. All symptomatic and all presymptomatic patients require lifelong decoppering with careful clinical tracking. Decoppering ensures that presymptomatic individuals remain symptom free. With judicious decoppering, given time, even patients with severe neurological disability improve and can return to normal life and resume school or work at par with their peers. Treatment regimens and tracking patients using the WD-specific Global Assessment Scale for WD (GAS for WD) are discussed.

KEYWORDS: ATP7B gene; Copper; Decoppering; Global Assessment Scale for WD (GAS for WD); Hepatocellular degeneration; Kayser–Fleischer rings; Penicillamine; Trientine; Wilson disease; Zinc

PMID: 24209444 DOI: 10.1016/B978-0-12-410502-7.00014-4

[Indexed for MEDLINE]