AAVBC
AMERICAN ACADEMY OF VALUE BASED CARE

Wilson's Disease

Quick Reference Guide

2026

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1 CLINICAL SNAPSHOT

Definition: Wilson disease (WD; hepatolenticular degeneration) is a rare autosomal recessive disorder caused by pathogenic variants in **ATP7B**, impairing biliary copper excretion and copper incorporation into ceruloplasmin.[1,2] Copper accumulates in the **liver, brain, cornea, kidneys, and other tissues**, producing hepatic, neurologic, psychiatric, ophthalmologic, and renal manifestations. WD is highly treatable when recognized early, but delayed diagnosis can lead to irreversible liver failure, neurologic disability, or death. For PCPs, the essential clinical point is simple: Wilson disease is difficult to differentiate; PCPs should maintain a low threshold for testing when hepatic, neurologic, or psychiatric findings are otherwise unexplained.[1,2]

ICD-10 Codes:

Confirmed Wilson disease is reported with **E83.01 — Wilson disease**. Avoid the nonspecific parent codes **E83.00 — Disorder of copper metabolism, unspecified** and **E83.09 — Other disorders of copper metabolism** once Wilson disease has been confirmed; under current CMS-HCC models, neither maps to an HCC, and both obscure the inherited metabolic diagnosis. Because Wilson disease is systemic, each active manifestation should be coded separately and linked to the underlying disease. Examples include **K74.60** for cirrhosis, **K72.10/K72.11** for chronic hepatic failure without/with coma, **R25.1** for tremor, **G25.3** for myoclonus, **G24.9** for dystonia, **G21.8** for secondary parkinsonism, **R27.0** for ataxia, **G25.5** for chorea, **F06.8** for a psychiatric disorder due to a known physiological condition, **H18.041** (right eye), **H18.042** (left eye), **H18.043** (bilateral), or **H18.049** (unspecified eye) for Kayser-Fleischer rings, **M81.0/M81.8** for osteoporosis, and **N04.9** for nephrotic syndrome when related to D-penicillamine toxicity (with the appropriate adverse-effect code also assigned when drug-induced). First-degree relatives undergoing evaluation may also carry **Z83.49** for family history of other endocrine, nutritional, or metabolic disease.[20] Documentation should state "**Wilson disease,**" "**Wilson's disease,**" **or** "**hepatolenticular degeneration**" and include the diagnostic basis, such as elevated 24-hour urinary copper, low ceruloplasmin, Kayser-Fleischer rings, increased hepatic copper concentration, or biallelic **ATP7B** pathogenic variants.[1] Use direct causal wording such as "**cirrhosis secondary to Wilson disease**" or "**tremor due to Wilson disease**" rather than listing manifestations as unrelated diagnoses.[1]

Prevalence and Burden: Traditional estimates place Wilson disease prevalence at approximately **1 in 30,000**, although molecular studies suggest that ATP7B-associated disease may occur as frequently as 1 in 7,000 to 1 in 20,000 individuals, with variable clinical penetrance.[1,2] Carrier frequency for a single pathogenic ATP7B variant is approximately **1 in 90**.[1,5] Most patients are diagnosed before age 40, but **older age does not exclude Wilson disease**.[1] Confirmed cases have been reported in patients in their 70s and 80s, and late-onset disease may present predominantly with neurologic or psychiatric findings rather than obvious hepatic disease.[1]

CLINICAL PEARL - TEST THE PATTER, NOT THE AGE

Wilson disease is often mistaken for more common hepatic, neurologic, or psychiatric disorders, and **psychiatric symptoms may precede liver or neurologic findings**. In any patient with otherwise unexplained liver disease, movement abnormalities, neuropsychiatric symptoms, or Coombs-negative hemolytic anemia, order **serum ceruloplasmin and 24-hour urine copper—regardless of age**. A **normal or low-normal ceruloplasmin does not exclude Wilson disease.**