EDITORIAL

Wilson's Disease in Children

A genetic illness called Wilson's disease (WD) occurs when the ATP7B protein fails, causing copper to build up in the liver, brain, and other organs. Patients typically present with liver disease, neuropsychiatric symptoms, ophthalmologic abnormalities, Kayser-Fleischer corneal rings.[1-3] Examples of hepatic disease include clinically asymptomatic cases with mild liver enzyme elevation and steatosis, as well as clinically overt advanced chronic liver disease (ACLD) with varices, splanchnic collaterals, splenomegaly, or hepatic decompensation. Fulminant hepatic failure (usually caused by ACLD) with Coombs' negative hemolytic anemia is rare.[4] In Bangladesh Shishu Hospital & Institute studies, 70% of patients were hepatic, 16.7% were neuropsychiatric, 5.0%–30% were hepatic and neuropsychiatric, and 8.3% were asymptomatic.[5] Another BSMMU study found that 22 (66.7%) individuals had hepatic symptoms and 10 (33.3%) had both.[6] Burghart and colleagues[7] found that 35.8% of 137 patients with Wilson's disease (Leipzig score ≥4) showed symptoms of CSPH: 14 (10.2%) had varices, 40 (29.2%) had splenomegaly, 20 (14.6%) had ascites, 18 (13.1%) had hepatic encephalopathy, and 3 (2.2%) had acute variceal hemorrhage. Kayser–Fleischer (KF) rings are bilateral and seen in 50–60% of patients with hepatic WD and 95–100% of patients with neurological WD. A study at BSMMU's Pediatric Gastroenterology and Nutrition Department found that the K-F ring was present in 84% of patients with only liver issues from Wilson's disease and in 90% of patients with neurological issues from Wilson's disease.[8] The practitioner must suspect and thoroughly explore the child with persistent asymptomatic elevated aminotransferase levels to diagnose and treat early.[9]

Unless genetic testing is done, no single laboratory test can validate this diagnosis without clinical and laboratory data. The group at the Eighth International Conference on Wilson Disease created the Leipzig Score in 2001, which uses lab tests (like serum ceruloplasmin, urinary copper excretion, and liver copper content) and clinical signs (such as neuropsychiatric symptoms, Kayser-Fleischer rings in the eyes, and Coombs' negative hemolysis) along with genetic testing. Total score: 4+—diagnosed; 2-3—possible; 0-1—very unlikely.[10] Repeated liver biopsy is impractical for liver disease tracking. Evaluation included non-invasive tests, platelet count (less than 150 × 109/L), and transient elastography (TE) for liver stiffness measurement (LSM).[11] Baveno VII guidelines suggest that compensated ACLD patients with LSM by TE < 15 kPa and platelet count ≥150 × 109/L are excluded from CSPH.[12] WD patients with LSM (TE < 15 kPa) and normal platelet count had a decreased ACLD progression risk and improved 10-year TFS. WD patients with ACLD and LSM ≥15 kPa showed decompensation and lower TFS.[13]

The first treatment relies on copper chelators like D-penicillamine and Trientine to reduce copper levels. Lifelong maintenance therapy with D-penicillamine or trientine or zinc is required. Starting with 250–500 mg/day of D-penicillamine and progressively increasing it by 250 mg every 4–7 days to 1000–1500 mg/day (15–20 mg/kg/day, up to 2000 mg/day) in 2–4 smaller doses may help patients tolerate the medicine. In adults, the maintenance dose is 10–15mgkg/day (775–1000 mgday) split twice. In youngsters, start with 20 mg/kg/day in two or three doses and gradually lower to 10–15 mg/kg for maintenance. Food reduces D-penicillamine absorption, therefore take it 1 to 2 hours before or after meals. D-penicillamine patients use 25–50mg of pyridoxine daily. Clinical and biochemical improvements and 24-hour urine copper excretion during treatment indicate therapeutic efficacy. This is highest shortly after treatment, sometimes exceeding 1000-2000 ìg/24 h. Urinary copper excretion should be 200–500 g/24 hours under chronic maintenance.[14] Patients who cannot tolerate D-penicillamine may take trientine. Trientine may be better for Wilson's disease patients with severe thrombocytopenia or neutropenia, which can cause splenomegaly and paradoxical neurological deterioration. First-line zinc treatment for asymptomatic patients is widespread.[15-17] Cu chelation therapy is uncertain even when started and maintained. Advanced cirrhosis and its complications can kill after a long life. Only an emergency liver transplant can save such

cases.[18] All siblings should be examined for Wilson disease since early identification and treatment improve outcomes.[19]

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