A clinical study of Wilson's disease: The experience of a single Egyptian Paediatric Hepatology Unit

Background and study aims: Most paediatric patients with Wilson's disease (WD) present with hepatic manifestations, but some may have neurologic or psychiatric features. Our aim was to define the clinical, biochemical features and the outcome of therapy of a group of Egyptian children diagnosed with WD. Patients and methods: The study was carried out at the Paediatric Hepatology Unit at Cairo University Children's Hospital, Egypt; 54 patients were diagnosed with WD from 1996 to 2009. The diagnosis was based on low serum ceruloplasmin levels, increased urinary copper concentrations before or after D-penicillamine challenge and/or the presence of Kayser–Fleischer (K–F) rings. Results: The clinical presentation was as follows: hepatic presentation in 33 patients (61%), hepato-neurologic 3 (5.5%), neurologic 5 (9.3%) and presymptomatic 13 (24%). Twelve couples had more than one affected sib. Increased urinary copper concentrations before or after D-penicillamine challenge was found in all patients, low serum ceruloplasmin in 97% and K–F rings in 31.5%. All patients were treated with penicillamine and zinc sulphate except one presymptomatic case who was treated with zinc sulphate only. Three patients underwent liver transplantation and eight patients died after a median duration of treatment of 6 months (1–36). The hepatic symptoms improved with treatment but the neurological symptoms remained stationary.

Conclusions: Clinical and biochemical assays remain the standard for diagnosis of WD. Penicillamine and zinc therapy can effectively treat WD with hepatic symptoms. Liver transplantation remains life saving for those with fulminant and end stage WD. Screening for presymptomatic sibs is of utmost importance.