CLINICAL and NEUROLOGICAL OF WILSON–KONOVALOV DISEASE IN CHILDREN

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Relevance. Wilson–Konovalov disease (WCD) is a hereditary disorder caused by mutations in the ATP7B gene, which is responsible for copper transport and metabolism in the body. Copper is a trace element essential for normal physiological function; however, mutations in the ATP7B gene lead to copper accumulation in various tissues and organs, resulting in their damage.

Objective. To study the clinical, neurological, and laboratory features of Wilson–Konovalov disease in children.

Materials and Methods. The study included 40 participants divided into two groups: the main group – 20 children with Wilson–Konovalov disease from consanguineous marriages, and the control group – 20 healthy children aged 5 to 14 years. Family history was assessed, cognitive functions were evaluated using neuropsychological scales (Wechsler scale), along with ophthalmological, biochemical (copper and ceruloplasmin levels in urine and blood), and instrumental (liver ultrasound) examinations.

Results and Discussion. Medical history revealed that in the main group, the first symptoms in all patients were general malaise (weakness, fatigue, recurrent epistaxis). In 7 children (33.3%) over the age of 10, neurological manifestations such as dysarthria, dysphagia, hypomimia, and affective disorders were identified. Cognitive assessment using the Wechsler scale showed moderate intellectual decline in 2 children (13.3%). Ophthalmological examinations, including slit-lamp evaluation, revealed Kayser–Fleischer rings in 4 children (26.7%). Daily urinary copper excretion (normally <40 µg/day) exceeded 90 µg/day in 6 children (40%) with clinical signs of WCD. Serum ceruloplasmin levels in the main group averaged 0.9±0.2 g/L (normal 0.2–0.4 g/L), while total serum copper was 9.8±1.1 µmol/L (normal 12.56–24.34 µmol/L). Moderately reduced ceruloplasmin was noted in 5 children (33.3%). Abdominal ultrasound revealed hepatomegaly and varying degrees of gallbladder enlargement.

Conclusions. The study was based on data from 40 children aged 5 to 14 years. The first complaints among children with WCD (main group) were rapid fatigability. It should be emphasized that neurological symptoms appeared after 8–10 years of age. Biochemical studies demonstrated decreased serum ceruloplasmin levels and elevated daily urinary copper excretion.