Clinical and laboratory characteristics of different stages of Wilson's disease

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Introduction: Wilson`s disease (WD) is a rare hereditary disorder of copper metabolism with excellent prognosis, if diagnosed on time. Search of the new simple diagnostic criteria is of great importance. Objective: To study the most frequent combinations of clinical and laboratory findings in patients with different stages of WD. Methods: Clinical, neurological, biochemical and haemostatic status was assessed in 22 patients with hepatic and in 53 patients with neurologic stage of WD. Results: Initial clinical manifestations were distributed as following: signs of liver affection – 43%, hemorrhagic syndrome (nasal, gingival or cutaneous) – 30%, primary neurologic symptoms – 16% and the other variants - 11%. Hyperkinesia, mixed dysarthria, hemorrhagic and menstrual disorders was the most typical combination in 73% of cases in the neurologic stage of WD. Hemorrhagic and menstrual disorders coupled with asthenic autonomic syndrome and mild action tremor in hands were identified in 65% of patients in the hepatic stage. Disturbances of copper metabolism (91%), cytolysis (59%) and cholestasis (52%), thrombocytopenia (57%), changes in platelet function (96%) and coagulopathy (60%) were the leading biochemical findings. Cytolysis and cholestasis were more common ($p = 0.04$ and $p = 0.03$, respectively) and more evident in patients with the hepatic stage, while coagulopathy and thrombocytopenia - in the neurologic stage of WD ($p = 0.03$ and $p = 0.02$, respectively). Conclusion: Different combination and severity of clinical and laboratory findings in patients with hepatic or neurologic stage of WD should be considered in diagnosing.