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Efficacy of D-penicillamine Challenge Test for Diagnosis of Wilson Disease

Norikazu Shimizu[1)], Atsuko Watanabe[1)], Junko Nakazato[1)], Hiroaki Nakamura[1)], Misako Inoue[1)], Kaoru Hirai[1)] and Tsugutoshi Aoki[1)]

1) Second Department of Pediatrics, Toho University School of Medicine

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Abstract:
Wilson disease is an autosomal recessive disorder of copper metabolism characterized by hepatic and/or neurological manifestations. This biochemical features of this disease are low serum ceruloplamine levels and high urinary copper excretion. Early diagnosis is very important to improve the prognosis of this disease. However, some patients revealed atypical biochemical findings. This study presents the efficacy of D-penicillamine challenge test for diagnosis of Wilson disease. Five patients and five normal controls were loaded 20mg/kg of D-penicillamine. Urinary copper / body weight(kg)ratio and/or urinary copper / creatinine ratio showed significant difference between Wilson disease patients and controls. The D-penicillamine challenge test will be useful for diagnosis of Wilson disease.

Key words: Wilson disease, inborn error of copper metabolism, D-penicillamine, D-penicillamine challenge test, urinary copper excretion

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