Different brain pathology in neurological and hepatic forms of Wilson disease depicted by quantitative MRI

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Purpose: Quantitative MRI is a sensitive tool to study microstructural changes in tissues. This study aims at comparison of differences in quantitative MRI metrics in the deep gray matter nuclei in Wilson disease (WD) patients with neurological and hepatic form. Subjects/Methods: 40 patients with genetically confirmed WD (30 neurological WD, 10 mild hepatic WD) on a stable anti-copper treatment and 26 healthy controls were investigated. 3T MR system was used to study quantitative susceptibility and T2/T1 relaxation times in globus pallidus, putamen, caudate nucleus and thalamus. Serum ceruloplasmin oxidase activity was measured using the o-dianysine assay. Results: T2 relaxation times were significantly lower in neuro-WD group in globus pallidus, putamen, and caudate nucleus compared to controls and hep-WD, whereas T2 relaxation times in hep-WD did not differ from controls. T1 relaxometry revealed higher T1 values in the thalamus in neuro-WD group compared to both controls and hep-WD patients (p0.02). Significantly higher susceptibility values were found in all studied regions in neuro-WD patients compared to controls and hep-WD patients (p0.02). No difference was found between hep-WD patients and controls. No correlation with the ceruloplasmin oxidase activity was found. Conclusion: Our results show that hep-WD is not associated with significant brain pathology. Decreased T2 relaxation time/ increased susceptibility in the basal ganglia indicate iron deposits, whereas increased T1 relaxation time/ increased susceptibility in the thalamus correspond rather to demyelination in neuro-WD group. Iron accumulation is not causally related to decreased ferroxidase activity. Supported by MH CR 15-25602A and 00023001IKEM Institutional support.