Wilson disease - neurological presentation

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Neurological presentation of Wilson disease (WD) is common, and it affects approximately 50% of patients. Neurological symptoms occur later compared to hepatic manifestations, typically in the 3rd or 4th decade. Most common symptoms include various combinations of movement disorders, namely tremor, dystonia, Parkinsonism, and chorea associated with dysarthria, dysphagia, and drooling. Damage to the central nervous system in WD is caused by the toxic effect of accumulated copper. It can be visualized as atrophy and increased signal of affected regions on T2 weighted magnetic resonance images. T2 hyperintensities may reflect tissue edema, demyelination, gliosis and rarefaction, whereby the latter two abnormalities are irreversible. The best treatment for neurologic symptoms is prevention; they do not occur when adequate lifelong anti-copper treatment is initiated in the asymptomatic or hepatic stage of the disease. Currently, there is no consensus about the optimal treatment of the neurologic manifestation and different WD centers favor Zinc, chelating agent, or combination of both. Neurological deterioration, sometimes irreversible, that is observed after the initiation of the treatment in 20-50% patients remains the most arduous problem in the WD treatment.