WILSON'S DISEASE PRESENTING WITH THE EPISODIC HYPERSOMNIA

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Case: A 48-year-old woman was admitted because of fluctuating cognitive impairment for 2 years. Her complaints as fatigue, decreased level of concentration, and depression started at 3 years ago. She developed behavioral changes, emotional lability, restlessness and agitation. There was no specific family history. No abnormality was found at physical and neurologic examination. A moderate elevation in liver function tests was found, but the all viral hepatitis markers and VDRL were negative. Neither toxic nor infectious disease was detectable. The initial diagnosis of WD was based on decreased ceruloplasmin level, increased urinary copper excretion, and decreased serum copper. Kayser-Fleischer (KF) ring was not detected. Brain MRI disclosed increase T1 signal in both globus pallidus. EEG showed diffuse slow waves without epileptiform discharge. Liver biopsy was performed at 3 years after symptoms onset and showed the presence of nuclear glycogen with cytoplasmic brownish pigment in hepatocytes and a minimal degree of periportal fibrosis, but no sign of hepatitis or other liver disease was detectable. EM changes included mitochondrial swelling and crista defragmentation. No evidence of hepatic encephalopathy was found. Blood ammonia level was normal. At 8-10 months after the initiation of the D-penicillamine therapy her complaints gradually resolved. In one year since resolution, she became drowsier and more hypersomnolent episodically. During these attacks she spent more than 2/3 of the day sleeping. After the administration of methylphenidate, she became alert and gradually improved negativism.

Conclusions: We present the Wilson's disease (WD) case with the episodic hypersomnia as the only symptom.