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Prevalence and features of non-motor symptoms in Wilson's disease

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Introduction: Wilson's disease (WD) is an autosomal recessive disorder caused by excessive copper deposition in liver, brain and other organs [1,2]. The clinical picture is characterized by hepatic, psychiatric and neurological dysfunction. Movement disorders are the core neurological features, although non-motor symptoms (NMS), as cognitive/affective, autonomic and sleep disorders, may occur over time [2,4]. We aimed to assess the frequency of NMS in WD patients compared with healthy subjects.

Methods: Twenty-seven patients affected with genetically proven WD (12 F, 15 M) and 35 healthy controls (Ctrl; 17 F, 18 M), comparable for age and education, were enrolled. Eighteen patients presented with the neurological form of the disease (NV) and nine with the non-neurological variant (NNV) [3]. NMS were assessed in all subjects by the following clinical scales: Mini-Mental State Examination (MMSE), Non-Motor Symptoms Scale (NMSS), SCOPA-AUT Questionnaire, Apathy Evaluation Scale (AES), Beck Depression Inventory (BDI), Epworth Sleepiness Scale (ESS), Restless Legs Syndrome Rating Scale (RLSRS), REM Sleep Behavior Disorder Screening Questionnaire (RBDSQ), Questionnaire for Impulsive-Compulsive Disorders in Parkinson's disease (QUIP-RS).

Results: We found that the patients showed more severe and frequent NMS and daytime sleepiness, and lower MMSE than Ctrl. In comparison to healthy subjects, NV subjects showed statistically significant higher ESS, NMSS, and RLSRS scores, and a lower MMSE score. Subtle and subclinical extrapyramidal/pyramidal signs and brain MRI signal abnormalities were detected in patients considered as asymptomatic for neurological disturbances.

Conclusions: NMS are common among WD patient, in particular those with NV, likely due to the widespread pathological changes throughout the central nervous system.

References:

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