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ARE PSYCHIATRIC SYMPTOMS A CORE PHENOTYPE OF MYOCLONUS DYSTONIA SYNDROME CAUSED BY SGCE MUTATIONS?

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Objective Myoclonus Dystonia Syndrome (MDS) is a childhood onset, alcohol responsive movement disorder caused by mutations in the SGCE gene in a proportion of cases. Single family and case series have suggested co-morbid psychiatric disease but have not compared cases to a control group.

Aims To establish a cohort of MDS patients with SGCE mutations and a control group of alcohol-responsive tremor patients, and to systematically assess for psychiatric symptoms using standardised questionnaires.

Method We collected 27 patients with SGCE mutations and 45 tremor control cases. The MINI International Neuropsychiatric Interview, PHQ-9, MADRS, YBOCS and AUDIT were used to assess psychiatric disease according to DSM-IV criteria.

Results There was a higher rate of psychiatric disease in MDS patients compared to controls ($p < 0.05$), specifically social phobia ($p < 0.05$) and Obsessive-Compulsive disease (OCD) ($p < 0.001$). Excess alcohol use was higher amongst the MDS group once cases $> < 18$ yrs were excluded. >

Conclusion Overall psychiatric disease is elevated amongst the MDS cohort compared to a control group with a chronic, socially stigmatizing disorder. OCD appears to be the greatest contributor to this effect and may reflect a pleiotropic function for the SGCE gene.