The hypocretin neurotransmission system in myotonic dystrophy type 1. Authors: Ciafaloni E, Mignot E, Sansone V, Hilbert JE, Lin L, Lin X, Liu LC, Pigeon WR, Perlis ML, Thornton CA.

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Excessive daytime sleepiness (EDS) is a common and often significant symptom for patients with myotonic dystrophy type 1 (DM1). It has been reported that narcolepsy-like sleep-onset REM has been observed in some patients with DM1. Similarly, the hypocretin (Hcrt) neurotransmission, which is impaired in narcolepsy, was reported previously to be reduced in DM1.

The current study further investigated the Hcrt neurotransmission system in DM1 for patients with and without EDS. Patients were assessed by the Epworth Sleepiness Scale (ESS) to measure symptoms of EDS. Lumbar puncture was performed to measure and analyze Hcrt values in spinal fluid. Participants from the University of Rochester Medical Center also underwent tests to measure their sleep patterns in the laboratory. These sleep exams included a polysomnography (PSG) and a Multiple Sleep Latency Test (MSLT).

Results indicated that Hcrt-1 values and Hcrt splicing in DM1 were not different compared to controls. Levels of Hcrt-1 were also similar between patients with DM1 who reported varying degrees of EDS symptoms on the Epworth Sleepiness Scale (ESS). Both Hcrt-1 level and ESS score were not found to be associated with the size of CTG repeat expansion. There were abnormal results observed during the sleep studies. Patients with DM1 had a short sleep-onset REM. The results of these sleep exams and laboratory tests helped the authors conclude that excessive sleepiness in DM1 is different from the sleep disturbance that occurs in narcolepsy.. More research is needed to determine the causes of excessive sleepiness in DM1 and the most appropriate therapies.

More results and conclusions can be found at:
http://www.ncbi.nlm.nih.gov/pubmed/18195268?ordinalpos=1&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_RVDocSum