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Title: Progressive weakness, ptosis and pseudomyopathic face as the presenting features in a patient with narcolepsy

Authors: JR, Fatemah, ?Albert, KA, RW, AB

Objective - To highlight how clinical signs common to myasthenia gravis (MG) can complicate the diagnosis of Narcolepsy with cataplexy

Method - Case Report

Results - A previously well 12 year old boy presented with a 2 month history of progressive bilateral weakness, ptosis and fatigability. Infrequent episodes of legs buckling were reported.

On examination his face appeared myopathic with bilateral ptosis. Hypophonia with slow speech was evident. Eye movements were normal without nystagmus. Fatigability was demonstrable on upwards gaze and repetitive shoulder abduction.

MRI head and spine were normal. EMG demonstrated jitter and blocking suggestive of a defect of neuromuscular transmission. Tensilon testing was not performed. Pyridostigmine was started and titrated over several days with reduced weakness and fatigability recorded. Ptosis and persistent tiredness were unchanged.

Five days into admission the patient had a witnessed episode of sudden collapse with loss of tone but retained consciousness brought on by laughing. A diagnosis of cataplexy with narcolepsy was made.

The patient was acetyl-choline receptor antibody negative, and positive for HLA DQB1*06:02. Pyridostigmine was stopped and methylphenidate started, which improved daytime sleepiness. Ptosis improved slightly but cataplexy persisted. Venlafaxine was introduced after 2 weeks, with clear improvement in ptosis, no further falls and return to school.

Conclusion - Narcolepsy with cataplexy typically presents as excessive daytime sleepiness with sudden onset bilateral loss of tone when experiencing strong emotions and may include hypnopompic/hypnogogic hallucinations and sleep paralysis. Prepubertal cataplexy can present with features mimicking the clinical signs of MG with fatigability, hypophonia and pseudo-myopathic face. The neurophysiological findings in our patient are intriguing. Investigation of the neuromuscular junction in other patients with narcolepsy is an interesting avenue for future work.