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Myasthenic or cataplectic facies? Ice pack test response in paediatric type 1 narcolepsy

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1. Introduction to the case

A eight-year-old boy presented with subacute onset of bilateral ptosis, droopy face and sleepiness. Basing on the former features, investigations were initially focused on a suspect of myasthenia gravis, that was also suggested by ice pack test result; this delayed further investigations. Sleepiness was initially overlooked, and only after three months the patient was referred for a polysomnography and multiple sleep latencies tests that revealed short sleep latencies and sleep onset REM periods. Low CSF hypocretin-1 level was consistent with type 1 narcolepsy (NT1). The patient was treated with sodium oxybate with resolution of ptosis, and benefit on sleepiness.

2. Image analysis

The ice pack test disclosed a bilateral ptosis resolution (Fig. 1), defined as an improvement of at least 2 mm in the distance between midpupil and the upper eyelid margin. This was different from myasthenia gravis, where limited positive response in the tested eyelid is expected. No sleepiness was reported by the patient during the test, as confirmed by simultaneous wake EEG.

3. Discussion

Classic features of cataplexy, such as relationship with emotions and falls, may be absent in childhood NT1 at presentation [1]. Semipermanent (fluctuating) eyelids weakness, typical of *cataplectic facies* in paediatric NT1, may mimic myasthenic disorders, potentially leading to misdiagnosis [1,2]. Additionally, sleepiness is often overlooked or misinterpreted as fatigue or inattentiveness, while hallucinations and nightmares may be mislabelled as psychological issues, further contributing to delay in diagnosis [3]. Consequently, NT1 patients are usually diagnosed 10–15 years after symptoms onset [4], resulting in detrimental effect on the children's learning and psychological development, weighting significantly on the already substantial burden that patients bear [5]. Cataplexy is thought to result from activation during wakefulness of the sleep circuitry involved in REM sleep that lead to reduced skeletal motor neuron activity via increased inhibitory and reduced excitatory signals [6]. Conversely, the cold stimulus benefit observed in disorders of the neuromuscular junction results from reduced acetylcholinesterase activity which leads to higher acetylcholine levels at the neuromuscular junction. We speculate that ice pack reversed the bilateral ptosis typical of the *cataplectic facies* via an activation of arousal and motivated behaviours under the control of the hypocretin/orexin system. Careful interpretation of ice pack test, as well as distinguishing among sleepiness and fatigability related to sustained effort, could help in the first evaluation, avoiding misinterpretation of the *cataplectic facies* [1], which often represents the challenging heralding picture of childhood NT1 [1].

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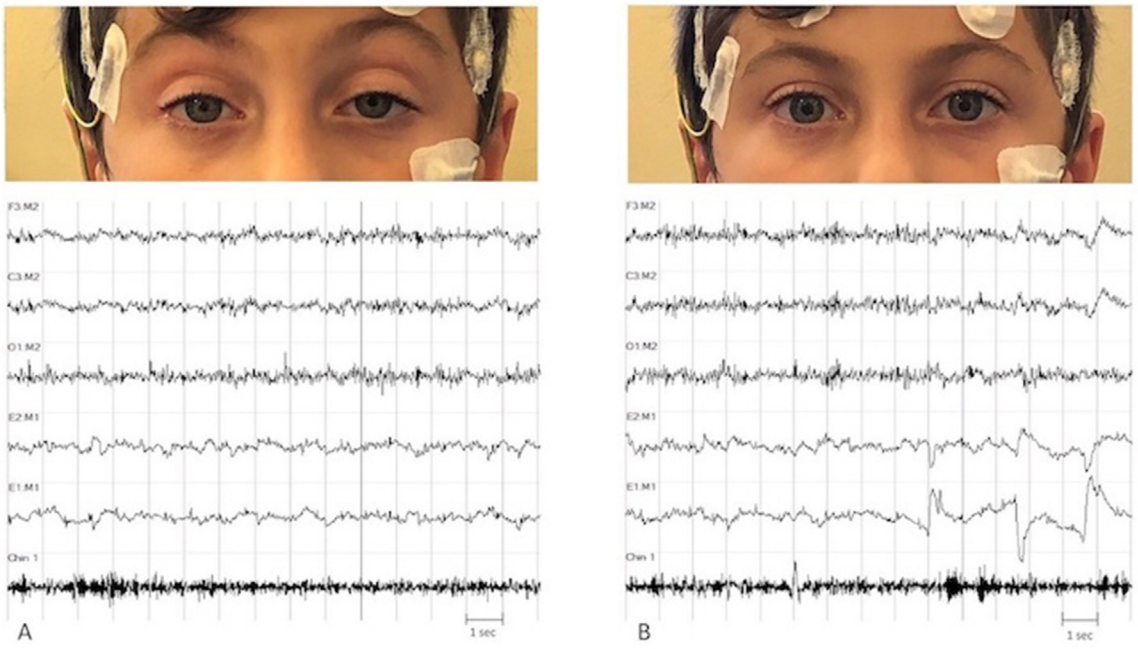

Fig. 1. A. Bilateral ptosis. EEG (F3-M2, C3-M2, O1-M2), electro-oculograms (E2-M1, E1-M1), chin EMG (Chin1) disclosed a normal wake pattern. **B.** Ice pack was applied on the left eyelid for 3-min resulting in bilateral resolution of ptosis without remarkable neurophysiological changes.

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Ethical compliance statement

The authors confirm that ethical approval from an institutional review board was not required. Written informed patient consent was obtained. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this work is consistent with those guidelines.

Informed consent

Written informed consent was obtained by patient parents.

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Conflict of interest

None.

The ICMJE Uniform Disclosure Form for Potential Conflicts of Interest associated with this article can be viewed by clicking on the following link: https://doi.org/10.1016/j.sleep.2021.06.045.

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