Case report

Nocturnal paroxysmal dystonia – Case report

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ABSTRACT

Nocturnal paroxysmal dystonia describes a syndrome consisting of recurrent motor episodes of dystonic–dyssynaptic features arising from non-rapid eye movement (NREM) sleep. In the article, the authors present a female case of nocturnal paroxysmal dystonia. The patient has had attacks since her childhood and was eventually diagnosed at the age of 48. Therapy with carbamazepine considerably reduced the frequency and extent of seizures. The present case evidences that nocturnal paroxysmal dystonia still is a diagnostic challenge for clinicians. Especially, we emphasize the importance of polysomnography in the verification of the diagnosis.

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1. Case report

This 48-year-old woman, was referred to the Department of Neurology, Charles Marcinkowski University of Medical Sciences in Poznań, Poland for further evaluation of episodes of involuntary movements occurring at night. She has had symptoms since her childhood, since she started walking. During those episodes her speech got slurred, involuntary movements of her face and limbs occurred, without neither enuresis, nor loss of consciousness or confusion after the attack. The episode lasted from a few seconds to 2 min, at a maximum, with the usual frequency of 4–5 seizures overnight. She remembered the episode in details and remained alert and in full logic contact through the attack. The patient complained of fragmentation of sleep, that was disrupted by frequent awakenings. Stress and fatigue made the attacks more severe and longer. Pregnancy (born 1 time, 2 miscarriages) made some relief of symptoms, but brought a few incidents of fainting. In the history, she revealed the chronic spinal pain syndrome and thyroid nodules. What is more in 1976 the patient underwent an appendectomy and in 1978 – cholecystectomy. In 2009, twice – in April and October – she was operated on hernia nucleus pulposus at levels C5-C6 and C6-C7. The patient was diagnosed by hematologist for thrombophilia, innate or acquired, with no certain patholgy found.

A family history of neurological diseases is negative. By admission, the patient was in good general condition, conscious, well oriented and logical, GCS ranged 15, with efficient cardio-respiratory system, and normal body temperature. Neurological examination at the admission showed a symptom of right-sided Hoffmann’s sign and lack of abdominal reflexes only. The laboratory tests, i.e. blood and urine tests,
brain MRI with the angio and venous program, EMG and interictal, video and Holter EEG – showed no abnormalities. Ophthalmologic examination was carried out with normal result.

During hospital stay the episodes of involuntary movements occurred every night, and – according to the patient – were identical to those reported earlier. They could be observed once during the daytime, while she was sleeping – the night before the patient had not slept getting ready for an EEG after a sleepless night. During the incident, dystonic movements of her limbs and facial dyskinesia were observed with the consciousness kept. During one of the incident blood samples were taken for electrolytes tests, i.e. sodium, potassium, calcium (also ionized), inorganic phosphorus and magnesium; all results were with normal range.

Carbamazepine treatment was implemented, at the initial dose of 100 mg, giving a significant reduction of seizures to about 2 overnight. With the dose of 200 mg, implemented on the very next day – there was a single episode, only. She began to sleep constantly through the night. At the discharge from the hospital the patient was recommended to take 300 mg of carbamazepine at night with further dose increasing under the control of neurologist.

Summing up – neurological examination, clinical picture with the film recorded by patient’s husband, the results of additional tests, and reasonably good response to carbamazepine, gave the reason for the diagnosis of paroxysmal nocturnal dystonia. In order to confirm the diagnosis polysomnography was performed. Polysomnographic (PSG) recording during one night showed 7 episodes of sudden, stereotyped movements, most often dystonic, with vocalization, lasting to 2 min. The attacks were preceded by abrupt awakenings, all the episodes occurred during NREM sleep stages N3 or N2. During PSG recording in the EEG trace no evident epileptiform discharges were recorded. Sleep architecture was characterized by reduced sleep efficiency to 70% due to increased amount of wake after sleep onset.

The patient, after the completion of diagnosis and treatment, was discharged from hospital in generally good condition.

2. Discussion

For the first time complex motor attacks with dystonic-dyskinetic movements were described in a group of 5 patients in 1981, by an Italian researchers Lugaresi and Cirignotta [1–4]. A few years later the condition was named nocturnal paroxysmal dystonia (NPD) [5].

Since then several cases of patients with paroxysmal nocturnal dystonia have been reported [5], however the syndrome still remains underestimated [5]. Paroxysmal nocturnal dystonia is a rare clinical entity characterized by the occurrence of motor episodes in the form of sudden movement – dyskinesia, most often dystonia, the symptoms occur during the sleep (night or day) and start at the stage of NREM sleep.

According to the International Classification of Sleep Disorders (ICSD, The International Classification of Sleep Disorders) in 2001 the disease was classified as other parasomnias [5]. In half of the patients the positive family history of parasomnias can be demonstrated and many patients present sleep disorders resembling parasomnias in their personal history [6].

Due to the incidental presence of the symptoms and signs and frequently-occurring bizarre symptoms the patients with NPD often remain, in general, underdiagnosed over many years or diagnosed with conversion disorder.

During the episode the dystonic movements are seen most often, often asymmetrical, but also of choreoathetoid, tonic or ballistic character [7,8] and may be accompanied by vocalization. What is diagnostically important – involuntary movements, even though, they may seem bizarre, are highly stereotyped [6]. Patients frequently remain fully conscious during the attack, that often awakes them at night. The incident is relatively short, lasting from 20–30 s to 1–2 min [7]. A third of the patients also experienced occasional generalized tonic-clonic seizures [9].

The incidents usually begin in childhood, they occur nearly every night, with the frequency ranged from a few to even several episodes during the night, resulting in significant deterioration of the quality of sleep and of life, as well.

Neurological examination is usually normal. In the most cases standard scalp EEG – ictal and interictal – usually remains unchanged or with no-specific changes [8]. This may be explained by activity of structures, that are not easily detectable by EEG surface electrodes [7]. Also, MRI picture usually presents no changes.

Kaido et al. reports that during ictal SPECT picture reveals hyperperfusion of the bilateral mesial frontal lobes, so ictal SPECT may be useful in the case of diagnostic doubts [3].

The Video-polysomnography demonstrating the stereotyped involuntary movements in NREM sleep is considered by many researchers as mandatory for the diagnosis, with full sensitivity and specificity [2,6]. However the polysomnography is not widely available and does not always capture the events, if they are not very frequent. But in most patients repeated video PSG can corroborate the final diagnosis.

The standard method of treatment is the administration of carbamazepine. The majority (approximately 70%) of patients stay on this treatment relatively well, even on small doses, the remaining 30% seems to be drug-resistant.

Nowadays more often NPD is considered to be an NREM sleep-related subtype of nocturnal frontal lobe epilepsy – NFLE [10]. This is because the partial seizures evoked from the additional motor cortex in the frontal lobe appear during sleep, being short-term and can occur without loss of consciousness [11]. The NFLE can include broad spectrum of symptoms: minor events – seizures lasting 2–4 s, paroxysmal arousals (PA) lasting 5–10 s associated with vocalization and sudden feeling of fear, and major attacks—the term for NPD [7].

NFLE also exists as an inherited form, more than a hundred families have been described [2]. The mutation on chromosome 20 and 15 was found [12], corresponding genetic defect of nicotinic acetylcholine receptor. However, it is still genetically heterogenous disorder and in some families those mutations cannot be established [5,6].
The differential diagnosis of NFLE should include sleep behavior disorders. The features that appeal more for NFLE than parasomnias are likely to be: attacks several times during the night, stereotype seizure, onset or persistence of symptoms into adulthood and involuntary movements of a tremor, dystonia or ballism [6].

In 2006 expert panel proposed Frontal Lobe Epilepsy and Parasomnia (FLEP) scale as a useful clinical tool in the differential diagnosis of movement disorders during sleep [13].

Extensive, diagnostic techniques like PET, SPECT and intracranial EEG may be promising in the confirming of the diagnosis, which often can register early focal seizures in the structures of the frontal and temporal lobes [3]. Paroxysmal nocturnal dystonia still remains an enigmatic disease with lots of questions to answer. Due to the small group of patients, less than 100 cases have been described [14], it is also difficult to develop diagnostic algorithm and standard therapy for patients not responding to carbamazepine.

In conclusion it is important to consider NPD in the differential diagnosis of nocturnal paroxysmal events as it is a disease that can be effectively treated in most cases.

Conflict of interest

None declared.

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Ethics

The work described in this article has been carried out in accordance with The Code of Ethics of the World Medical Association (Declaration of Helsinki) for experiments involving humans; Uniform Requirements for manuscripts submitted to Biomedical Journals.

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