



Narcolepsy and cataplexy: a pediatric case report

Tülin Savaş¹, İlknur Erol¹, Semra Saygı¹, Mehmet Ali Habeşoğlu²

¹Clinic of Pediatric Neurology, Başkent University School of Medicine Hospital, Adana, Turkey

²Clinic of Pulmonology, Başkent University School of Medicine Hospital, Adana, Turkey

Abstract

Narcolepsy is characterized by excessive sleepiness, cataplexy, hypnagogic hallucinations, and sleep paralysis during the rapid eye movement period of sleep. Herein, we present a boy aged eight years who was diagnosed as having narcolepsy and cataplexy about thirteen months after his first presentation. He was admitted with symptoms of daytime sleepiness. In the follow-up, cataplexy in the form of head dropping attacks developed seven months after the first admission. The patient was investigated for different prediagnoses and was eventually diagnosed as having narcolepsy and cataplexy through polysomnography and multiple sleep latency tests thirteen months after the first presentation. He is being followed up and is under drug therapy; his symptoms have improved substantially. (Turk Pediatri Ars 2016; 51: 221-3)

Keywords: Cataplexy, child, narcolepsy

Introduction

Narcolepsy is a rare disease caused by sleep disorder observed during the rapid eye movement (REM) phase of sleep, which is characterized by excessive sleepiness during daytime accompanied by sudden loss of muscle tone, hallucinations while falling asleep, and sleep paralysis (1). In children, the frequency has been estimated to be approximately 1/6 000 (2). It is thought to be related with disruption in hypocretin secretion as a result of autoimmune damage in cells that secrete hypocretin (orexin) in the lateral hypothalamus. A hypocretin level of less than 110 pg/mL in the cerebrospinal fluid (CSF) is diagnostic (3). In the diagnosis, multiple sleep latency test (MSLT) and CSF hypocretin measurement are needed in addition to appropriate clinical history. Presence of HLA DQB1 0602 allele strongly supports the diagnosis (2, 3).

Case

A boy aged 8 years presented with symptoms of excessive sleepiness, imbalance, and falling asleep rapidly. In his history, it was learned that he had had headache, malaise, blurred vision, and visual and auditory hallucinations

while falling asleep for the last one week, and he had no symptoms of fever or vomiting, and no history of any recent vaccination. There was no pathology in his personal and familial history. On systemic and neurologic examination, slow speaking, weak appearance, mild myosis in the pupils, and mild ataxia while walking in a straight line were observed. He was hospitalized with the prediagnosis of meningoencephalitis and acute disseminated encephalomyelitis (ADEM) and protein, glucose, cell count, oligoclonal band, and herpes simplex type 1 polymerase chain reaction were studied in the CSF. Brain and spinal magnetic resonance imaging (MRI), electroencephalography (EEG), and serologic tests for Lyme, Salmonella and Brucella were performed. These tests were found as normal and the patient was discharged after making an appointment with the division of pediatric psychiatry because of the presence of hallucinations and accompanying depressive findings. Seven months after the first admission, the patient presented again after developing instant relaxation during laughing and head drops, in addition to his earlier symptoms. He was investigated with the prediagnosis of epilepsy, subacute sclerosing panencephalitis (SSPE) and autoimmune encephalitis. Autoimmune antibodies (Anti GAD, Anti Hu, Anti Yu, Anti NMDA, voltage-gated

Address for Correspondence: Tülin Savaş E-mail: tulin_savas@hotmail.com

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K channel antibody) and CSF measles IgG antibody were negative. A video EEG examination was found normal and an appointment for polysomnography was made with a prediagnosis of narcolepsy. The patient was followed up in an external center for five months and he presented again when his symptoms continued to worsen. On physical examination, it was found that his body weight increased by 10 kg after the first presentation. Polysomnography and MSLT were performed 13 months after the first presentation. In the polysomnography, the total sleep time was 433 minutes, the time to fall asleep was 0.4 minutes, and the time to enter REM sleep was 4 minutes (Table 1). In the multiple sleep latency test, the mean time to fall asleep was 1.5 minutes and all four tests revealed that sleep began with REM sleep, so called sleep-onset rapid eye movement (SOREM) (Table 2). HLA DQB1*0602 allele, which was studied to support the diagnosis of narcolepsy was found positive. Modafinil and clomipramine treatment was initiated with a diagnosis of narcolepsy and cataplexy. In the follow-up visit performed one month later, it was learned that his symptoms had regressed markedly, sleepiness during the daytime was improved completely, cataplexy occurred rarely when he was extremely excited, he had started to attend school, and his academic success was good. On the follow-up examination, it was observed that his speech was more fluent and he lost weight. The patient is still being followed up in our clinic with a diagnosis of narcolepsy and cataplexy, and is receiving modafinil and

clomipramine treatment without any problems. Written informed consent was obtained from the patient's family.

Discussion

Although narcolepsy is known to be a condition that occurs in adults, it has been reported that the disease onset occurs in childhood and adolescence in most cases (4). In a retrospective study, it was found that the average time period between the onset of symptoms and the diagnosis was longer than 10 years in patients with narcolepsy (5). Patients with narcolepsy are usually initially investigated with the prediagnoses of epilepsy, encephalopathy, and psychiatric disease (5). Meningoencephalitis and acute disseminated encephalomyelitis were primarily considered in our patient because of sleepiness and slurred speech; investigations were performed and these prediagnoses were excluded.

Cataplexy is sudden loss of muscle tonus. It generally occurs with emotional responses including laughing, excitement, anger, and astonishment. It is observed in two thirds of patients with narcolepsy. Regional muscle involvement may be observed in these patients, as well as diffuse muscle involvement. The most commonly involved muscles include the muscles of the chin, neck, arms, and legs. During a cataplexy episode, drooping of the jaw or entire head, loosening of the arms on both sides, and twisting or loosening of the legs may be observed. This status may be confused with atonic seizures or negative myoclonus (6). In our case, cataplexy was primarily interpreted as negative myoclonus and therefore, the patient was investigated in terms of SSPE and atonic seizure. These prediagnoses were excluded by way of long-term video EEG monitoring and measles-specific IgG in CSF. Patients with cataplexy have been followed up with different prediagnoses in the literature, similar to our patient (7). A pediatric patient with local cataplexy was followed up with chronic symptoms of jaw dropping, jaw surgery was planned but the patient was diagnosed as having cataplexy a short time before surgery. Another patient was followed up with a diagnosis of conversion for many years (7).

The diagnosis of narcolepsy is supported by polysomnography and later by MSLT. A dozing-off time shorter than 15 minutes, frequently shorter than 5 minutes during the test and initiation of sleep during the REM period (achievement SOREM) are diagnostic (8). A diagnosis of narcolepsy was made in our patient with a mean dozing-off time of 1.5 minutes in the MSLT test and because each drowsing period of four initiated in REM sleep. In addition, sleep apnea syndrome was excluded by specifying that the sleep apnea index was 2 in polysomnography. Hypocretin is a

Table 1. Polysomnography findings of the patient

| | |
|-----------------------------------|------|
| Total sleep time (min) | 433 |
| Sleep efficiency (%) | 95.7 |
| Sleep latency (min) | 0.4 |
| REM latency (min) | 4 |
| Stage 1 sleep rate (%)*a | 5.8 |
| Stage 2 sleep rate (%)*a | 39.7 |
| Stage 3 sleep rate (%)*a | 37.8 |
| REM rate (%)*a | 16.7 |
| Apnea hypopnea index (event/hour) | 2 |

*Percentage by total sleep time

Table 2. Multiple sleep latency test results

| Test | Sleep latency | SOREM |
|------|---------------|-------|
| 1 | 30 s | Yes |
| 2 | 90 s | Yes |
| 3 | 120 s | Yes |
| 4 | 120 s | Yes |
| Mean | 90 s | |

SOREM: sleep onset rapid eye movement

recently-defined neuropeptide and its levels in CSF have been found low in approximately 90% of patients with idiopathic narcolepsy and cataplexy (8). We could not study the CSF hypocretin level in our patient because we could not find any laboratory that studied hypocretin levels, which is substantially important in making the diagnosis. In our country, CSF hypocretin level is not a routine test and as far as we know, no laboratories are testing hypocretin.

In most patients with narcolepsy, the DQB1 0602 allele is found as positive (3). Therefore, a positive DQB1 0602 allele in the presence of clinical findings is strong evidence for the diagnosis (3). In our case, the fact that the DQB1 0602 allele was found as positive supported our diagnosis.

Obesity frequently accompanies pediatric narcolepsy. This is thought to be related with accompanying hypothalamic dysfunction, excessive sleepiness during the day, and reduced school attendance (3, 8). In the literature, it was reported that a patient who had been followed up because of hypothyroidism-related obesity was diagnosed as having narcolepsy during the follow-up, and lost weight with appropriate treatment (7). In our patient, a 10 kg increase in body weight was found in a period of approximately one year, and normal body mass index was achieved after treatment. Therefore, body weight monitoring of children with narcolepsy during follow-up, as well as at the time of diagnosis is important in terms of the observation of treatment efficiency.

Although non-medical recommendations are present for the treatment of this disease (short naps during the day; exercise; diet recommendations; attention to sleep hygiene, which means to sleep at the same time each day and waking up at a certain time in the morning; achievement of better quality sleep by way of isolation of bedroom in terms of heat, noise, and light, and using appropriate beds; it has been recommended that medical treatment should be initiated immediately after the diagnosis is made and continued lifelong considering worse and painful outcomes of the disease in children and adolescents (6, 9). Modafinil and imipramine treatment was initiated in our patient after the diagnosis was made. Our patient is still being followed up with modafinil and imipramine treatment and his symptoms have markedly decreased.

In conclusion, narcolepsy is accompanied by learning difficulties and obesity because its symptoms are distributed in a wide spectrum, which may suggest many differ-

ent conditions, and affected children are often labeled as lazy. It is substantially difficult to make the diagnosis in patients with mild symptoms and patients are followed up with different prediagnoses in different outpatient clinics. The diagnosis is made years after the onset of symptoms in most cases. The fact that few pediatric cases have been reported in our country suggests that the diagnosis is being missed in this group of patients. Our patient was diagnosed 13 months after presentation and we presented this case in order to draw attention to narcolepsy and cataplexy in pediatric patients.

Informed Consent: Written informed consent was obtained from the patient's parents.

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