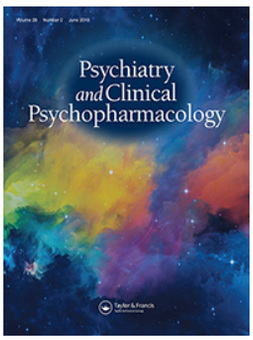

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CASE REPORT

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Quickly diagnosed and treated prepubertal Type 1 narcolepsy case

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ABSTRACT

Excessive daytime sleepiness, cataplexy, sleep paralysis, and hypnagogic hallucination are the classic tetrad of narcolepsy. It has been shown that narcolepsy, a chronic and disabling disease, starts in childhood and adolescence rather than adulthood. The International Classification of Sleep Disorder (ICSD-3) classifies narcolepsy into Type 1 (narcolepsy with cataplexy) and Type 2 (narcolepsy without cataplexy). There is low awareness and knowledge of narcolepsy among the general public, primary care physicians, and sleep specialists. It has been shown that the lack of recognition of disease symptoms delayed the diagnosis of narcolepsy from 8.7 to 22.1 years. In this case report, we will discuss the case of Type 1 narcolepsy, which started in the prepubertal period and was diagnosed and treated in a short period of time.

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KEYWORDS
Cataplexy; narcolepsy; prepubertal; sleep disorders

Introduction

Yoss and Doly [1] described the narcolepsy as a sudden and excessive daytime sleepiness (EDS), cataplexy, sleep paralysis, and hypnagogic hallucinations. DSM-5 criteria require EDS in association with any one of the following: cataplexy; cerebrospinal fluid (CSF) hypocretin deficiency; rapid eye movement (REM) sleep latency ≤15 min on nocturnal polysomnography (PSG); or mean sleep latency ≤8 min on multiple sleep latency testing (MSLT) with ≥2 sleep-onset REM sleep periods (SOREMPs) [2]. But only 10% of patients have all the classical narcoleptic tetrad. Automatic behaviours, poor focus, memory problems, and impaired night sleep are other common symptoms [3].

Type 1 narcolepsy (narcolepsy with cataplexy) is estimated to have a prevalence of 25–50 per 100,000 people and an incidence of 0.74 per 100,000 person-years [4]. The prevalence of narcolepsy in the paediatric population remains unknown, but based upon the regional study conducted in the U.S.A., it could affect 20–50 per 100,000 children [5]. Two peaks of onset are suggested, at ages 15–25 years and ages 30–35 years [2]. Cataplexy is pathognomonic for narcolepsy. Cataplexy is defined as reversible, sudden, striped muscle tone reduction, or loss triggered by emotional stimuli such as astonishment, fear, and laughter. There is no loss of consciousness during cataplexy attacks [6]. The International Classification of Sleep Disorder [7] (ICSD-3) classifies narcolepsy into Type 1 (narcolepsy with cataplexy) and Type 2 (narcolepsy without cataplexy). Cataplexy can develop in 10% of Type 2 narcoleptic patients and can be followed up with Type 1 narcolepsy.

In the pathophysiology of Type 1 narcolepsy, the loss of 95% of the neurons that produce orexin (hypocretin) in the lateral hypothalamus plays a role, which leads to the detection of low orexin levels in patient's CSF [8]. Narcolepsy is also associated with a low level of histamine in CSF that plays a role in wakefulness [9]. Narcolepsy is also known to be triggered by streptococcal upper respiratory tract infections [10]. In addition, recent years have seen the emergence of narcolepsy following influenza infections due to H1N1 or H1N1 vaccination [11,12].

Case presentation

A 9-year-old male patient was referred to child and adolescent psychiatry outpatient clinic because of involuntary sleep episodes during the day and sleeping a lot. The patient's symptoms first started 3 months ago. Two months ago, they presented to the paediatric neurology clinic for this reason. All the tests that had done in paediatric neurology were non-remarkable. Meanwhile, the amount of sleep in the patient began to increase. He also slept in school during lessons. Lately, he has been sleeping at the dinner table while eating. In this period, nervousness and aggressiveness, especially against family members, started. Within 2 months, his weight increased from 34 kg to 41 kg. The teachers of the patient complained that he had fallen asleep in

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the course, and at that time the school success of patient had regressed.

A brief medical history of the patient reveals that he was born in a normal, spontaneous vaginal way. There were no complications at birth. He had been breastfed for 18 months. Psychomotor development was usual. He used sodium valproate treatment with epilepsy diagnosis from 1.5 to 3.5 years. Characteristics of his seizures were generalized tonic clonic. He has not used any antiepileptic medications since he was 3.5 years old and has never had any epileptic seizures. There was no other medical illness. He was in the fourth grade and his school success was defined as medium; he is a child with good friendship relations and is loved by his friends. In the family history, there was no kinship between the parents. He has one sibling. He has a 4-year-old sister. There was no history of any psychiatric disorders in his family. No history of sleep disorders was found in the first- and second-degree relatives.

At the first visit with the patient, the patient had a general appearance of sleepiness. He was humming and giving short answers to the questions asked. The child was distracted during the interview, and many times he was drowsy. Despite being awakened, he fell asleep again in a very short time. The magnetic resonance imaging (MRI) of the brain, EEG, complete blood count, biochemistry (liver function tests, renal profile tests, chemistry panel, iron and iron binding capacity tests) and hormones (thyroid function tests, vitamin B12, and folic acid) were requested from the patient. The results were normal. The paediatric neurologist requested WISC-R to exclude mental retardation; because the patient did not understand the questions asked during the interview, gave delayed responses and the apathetic appearance. In the requested WISC-R result, the verbal intelligence score was 107, the performance score was 92, and the total intelligence score was 100. An endocrinology consultation was requested due to rapid weight gain to rule out diabetes mellitus. OGTT was performed and no endocrine abnormalities were found in their follow-ups. Result of PSG and MSLT with narcolepsy pre-diagnosis was reported as

> It was observed that the patient entered REM sleep 3 minutes after being taken to the room for recording. In MSLT shots performed 4 times in daytime with MSLT, REM start was detected in three of the shots, and it was evaluated in agreement with narcolepsy.

The patient diagnosed with narcolepsy as a result of PSG test and the patient was prescribed 25 mg imipramine by an adult neurologist as we prescribed short-acting methylphenidate. Behavioural treatment (includes developing healthy sleep habits and avoiding sleep deprivation) approaches were also recommended in the follow-up, and the patient's multiple sleeping and involuntary sleep episodes were regressed. The school success has been improved as it was before the disease. After the treatment, the patient's weight gain stopped and showed regression. It became clear that the patient's diagnosis was Type 1 narcolepsy (cataplexy narcolepsy) because the accumulation triggered by fear, excitement, and laughter a year after the narcolepsy diagnosis of the patient who stopped to gain weight.

Informed consent was received from the family.

Discussion

Narcolepsy is a chronic and disabling disease. It has been shown that narcolepsy begins in childhood and adolescence rather than adulthood [13]. It has been shown that the lack of recognition of disease symptoms delayed the diagnosis of narcolepsy from 8.7 to 22.1 years [14].

For the diagnosis of narcolepsy, it is essential that daytime sleepiness is present. The ICSD-3 [7] classifies narcolepsy into Type 1 (narcolepsy with cataplexy) and Type 2 (narcolepsy without cataplexy). PSG and MSLT were requested for the diagnosis of narcolepsy as the patient had EDS. With PSG, causes such as sleeping breathing disorders, which are more frequent in daytime extreme sleepiness, are excluded. MSLT is a sleep test applied after PSG in which at least 6 hours of sleep recording is performed, in the form of 4–6 episodes lasting 20 min each, and each episode is applied consecutively in 2-h intervals. It is a method that allows the measurement of sleeping speed and the detection of REM-initiated sleep [15]. Our patient entered REM sleep 3 min later. In addition, REM onset was found in three of the four MSLT shots. One year after the diagnosis of narcolepsy, it became clear that the diagnosis of Type 1 narcolepsy (cataplexy with narcolepsy) was based on the development of cataplexy in the patient. Since CSF sample was not taken from the patient, CSF hypokretin-1 level could not be measured.

Overall, 25% of children have had at least one sleep problem by adolescence, and this proportion can be as high as 75% in children with autism spectrum disorder, attention deficit hyperactivity disorder (ADHD), epilepsy, or headache [16]. At the same time, other sleep disorders (idiopathic hypersomnia, hypersomnolence disorder, Klein–Levine syndrome, etc.) should also be considered in the differential diagnosis of narcolepsy.

Obesity affects more than half of childhood narcoleptic cases [17]. Paediatric narcolepsy-cataplexy is also associated with excessive weight gain prior to symptom onset, and may be associated with other sleep disorders, including periodic limb movements and sleep apnoea, which may obscure or delay the correct clinical diagnosis [18]. Children with narcolepsy so appear to be overweight and/or obese [19], and an association with precocious puberty was anecdotally reported, suggesting the presence of a wider metabolic/hormonal

derangement [20]. The associated hypothalamic dysfunction is thought to be related to EDS and reduced school attendance [21]. In our patient, rapid weight gain after treatment stopped and started to regress afterwards. For this reason, it is important to follow the weight of the narcoleptic children in the follow-up to evaluate the effectiveness of the treatment, not only during the diagnosis. Hyperactive/aggressive behaviour in children, problems in communication with their peers, and rarely psychotic symptoms can be observed [22]. In our patient, improvement was observed in aggressive and nervous behaviours especially towards the family after treatment.

Surprisingly, with the sleep specialists, primary care physicians, and the general public who participated in the AWAKEN study [23], it was found that knowledge and awareness of narcolepsy was very low. It is important to identify the signs of narcolepsy early, to confirm the diagnosis and to make the treatment effective [22]. It is important to remember the diagnosis of narcolepsy in childhood patients who present with symptoms of daytime sleeping and excessive sleepiness, without forgetting that narcolepsy symptoms begin most often in childhood and adolescence.

Disclosure Statement

No potential conflict of interest was reported by the authors.

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