online©ML Comm

CASE REPORT

Psychiatry Investig 2007;4:121-123

Print ISSN 1738-3684 / On-line ISSN 1976-3026

Case Study of a Narcoleptic Patient with a Family History of Narcolepsy

Yoon-Kyung Shin, MD, Seung-Chul Hong, MD, PhD, Youn-Jung Cho, MD, Jong-Hyun Jeong, MD, PhD, Jin-Hee Han, MD, PhD, Sung-Pil Lee, MD, PhD

Department of Neuropsychiatry,
St. Vincent's Hospital,
The Catholic University of Korea
College of Medicine, Suwon, Korea

Narcolepsy is characterized by excessive daytime sleepiness, and this is typically associated with cataplexy and other rapid eye movement (REM) sleep phenomena such as sleep paralysis and hypnagogic hallucinations. These narcoleptic symptoms have been occasionally misdiagnosed as depression, seizure or even schizophrenia. The female patient (age=22 years) introduced here had experienced excessive daytime sleepiness, severe cataplexy and sleep paralysis. However, she didn't know the cause of her symptoms in spite of having visited hospitals several times for five years. Her father had also suffered from excessive daytime sleepiness since his early twenties. He had been diagnosed with depression and then took antidepressants; however, his sleepiness and fatigue didn't improve. This case history focuses on the diagnostic and therapeutic aspects of a patient with familial excessive daytime sleepiness.

KEY WORDS: Narcolepsy, Family, Excessive somnolence, Cataplexy.

Psychiatry Investig 2007;4:121-123

Introduction

Narcolepsy is a serious chronic sleep disorder that's characterized by excessive daytime sleepiness, sleep attacks and rapid eye movement (REM) sleep abnormalities (cataplexy, sleep paralysis and hypnagogic hallucination).[1] However, narcolepsy has only recently come under the focus of medical research. The symptoms of narcolepsy have often been mistaken for those of other disorders, for example, the excessive daytime sleepiness has been mistaken for idiopathic hypersomnia or for the fatigue of atypical depression, the cataplexy for epilepsy, and the vivid hypnagogic hallucinations for true hallucinations of a psychotic disorder. A family history of narcolepsy has been reported for 6% to 40% of narcoleptic individuals.[2-5] The risk of narcolepsy with cataplexy is estimated to be 10 to 40 times higher among the first-degree relatives of narcoleptic individuals than that for the general population.[6] The studies on the prevalence of narcolepsy in probands and their relatives have reported that 20% of patients with narcolepsy were familial cases.[1,7]

The patient of our current study, father and daughter all suffered from typical narcolepsy with excessive daytime sleepiness, sleep attacks and cataplexy, but they were not properly diagnosed for many years. This report introduces a case of typical familial narcolepsy.

Patient Description

A 22-year-old woman consulted with our department because of her excessive daytime sleepiness and sleep attacks. Her daytime sleepiness began when she was around thirteen years old and it had been getting worse. When she was in high school, she couldn't stay wake during her classes or exams. When she worked at a department store as a saleswoman after graduating from high school, she frequently dozed off

Correspondence
Seung-Chul Hong, MD, PhD
Department of Psychiatry,
St. Vincent's Hospital,
The Catholic University of Korea
College of Medicine,
93-6 Ji-dong, Paldal-gu, Suwon,
442-723 Korea
Tel +82-31-249-7161
Fax +82-31-248-6758
E-mail hscjohn@hotmail.com

standing up, and for this she was dismissed. She also got into a collision while driving due to a sudden sleep attack. Cataplexy developed as her daytime sleepiness got worse, and after the age of nineteen, it would occur almost every day. Cataplexy occurred when she laughed, got angry or exchanged jokes. It even occurred when she was excited, surprised and embarrassed, and when she reminisced about happy moments. Her knees would suddenly buckle and her jaws sagged. She also complained of seeing ghosts or animals and hearing her name called when she was lying down at night. She experienced realistic and often scary dreams throughout the night; when she awoke, she was unable to move.

There was no history of trauma to the head or any psychiatric illnesses. However, her father did suffer from constant sleepiness and fatigue, and also occasionally sudden muscle weakness of both knees when laughing. Although he consulted various hospitals, they told him nothing was out of the ordinary, but they recommended that he see a psychiatrist. The psychiatrist prescribed him 37.5 mg of venlafaxine and 0.75 mg of alprazolam for depression.

TABLE 1. The daughter's and father's clinical and polysomnographic characteristics

	The daughter	The father
Symptoms at interview		
ESS, score	19	18
Cataplexy	Several times a day	One or two a month
Sleep paralysis	Nearly every night	No
Hypnagogic hallucination	Nearly every night	No
MSLT		
Mean sleep latency	1.9 minutes	5 minutes
Number of SOREMPs	5	5
HLA-DQB1*0602	+	+

ESS: Epworth sleepiness scale, MSLT: mean sleep latency test, SOREMP: sleep-onset rapid eye movement (REM) period

However, he was still sleepy and languid in spite of those medications.

Both the father and the daughter underwent a multiple sleep latency test (MSLT) and HLA-DQB1*0602 allele typing. In addition, only the daughter underwent nocturnal polysomnography (PSG). On her nocturnal PSG test, her total sleep time was 429 minutes, and her awake time during sleep was 43 minutes. Her sleep efficiency was 91%. Her sleep latency and REM latency were very short at 1.3 minutes and 9.5 minutes, respectively. There were no respiratory disturbances, abnormal REM movements or behaviors during sleep. On MSLT of the daughter, the mean sleep latency was 1.9 minutes and sleep-onset REM periods (SOREMPs) occurred four times (Figure 1). On the father's MLST, the mean sleep latency was 5 minutes and SOREMPs occurred 4 times (Figure 2). The results of the HLA-DQB1*0602 typing for both patients were positive.

Both the father and the daughter were prescribed with 200 mg of modafinil and 75 mg of venlafaxine, and this resulted in reducing both their excessive daytime sleepiness and cataplexy, and especially for the father.

Discussion

This is a case of discovering the family connection of the patient who complained of excessive daytime sleepiness. Both the patient and her father suffered from cataplexy and on their MSLT, they showed sleep latency within 5 minutes and four SOREMPs. In conclusion, both of them met the criteria of the International Classification of Sleep Disorders (ICSD-2) for narcolepsy with cataplexy.[9] The HLA-DQB1*0602 allele results were positive for both cases, demonstrating a genetic influence. These two patients are an example of a more rarer case of familial narcolepsy in comparison with the usual non-familial narcolepsy.[2-4]

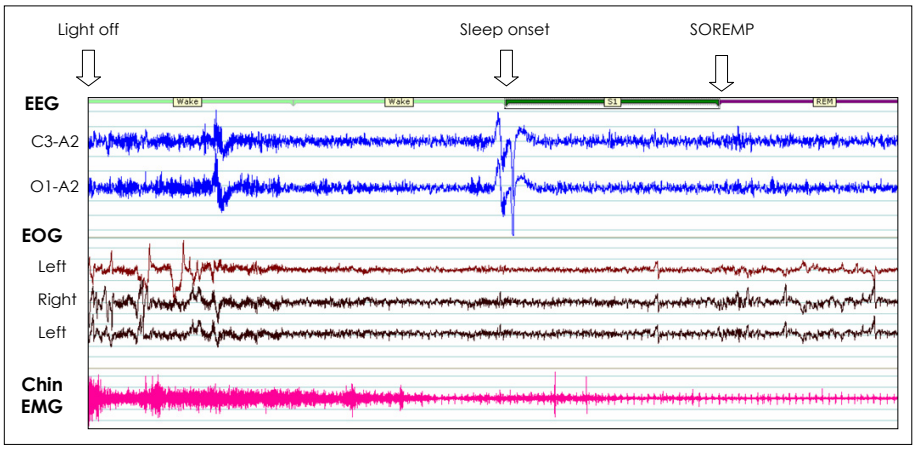

FIGURE 1. This figure demonstrates the sleep-onset REM period (SOREMP) of the daughter's mean sleep latency test (MSLT). Rapid eye movement (REM) began within 30 seconds of sleep onset, EEG: electroencephalogram, EOG: electrooculogram. EMG: electromyography.

According to Ohayon, 20 out of 96 narcoleptic probands (20.8%) were familial narcoleptics and the rest were sporadic cases.[1] In Billiard's study, there are 37 familial narcoleptics out of 188 probands (19.6%), which supported Ohayon's results.[7] The sooner narcolepsy develops and the greater the tendency for a familial connection, the worse is the degree of the symptoms.[8] As this case demonstrates, the symptoms of the daughter were severer and showed less response to medication than those of her father.

The symptoms of narcolepsy have often been mistaken for those of other disorders. Therefore, a physician must bear in mind of the possibility of sleep disorders, including narcolepsy, when faced with a patient complaining of excessive daytime sleepiness or fatigue.

REFERENCES

1. Ohayon MM, Okun ML. Occurrence of sleep disorders in the families of narcoleptic patients. Neurology 2006;67:703-705.
2. Nevsimalova S, Mignot E, Sonka K, Arrigoni JL. Familial aspects of narcolepsy-cataplexy in the Czech Republic. Sleep 1997;20:1021-1026.
3. Hayduk R, Flodman P, Spence MA, Erman MK, Mitler MM. HLA haplotypes, polysomnography, and pedigrees in a case series of patients with narcolepsy. Sleep 1997;20:850-857.
4. Guilleminault C, Mignot E, Grumet FC. Familial patterns of narcolepsy. Lancet 1989;2:1376-1379.
5. Baraitser M, Parkes JD. Genetic study of narcoleptic syndrome. J Med Genet 1978;15:254-259.
6. Nevsimalova S, Mignot E, Sonka K, Arrigoni JL. Familial aspects of narcolepsy-cataplexy in the Czech Republic. Sleep 1997;20:1021-1026.
7. Billiard M, Pasquié-Magnetto V, Heckman M, Carlander B, Besset A, Zachariev Z, et al. Family studies in narcolepsy. Sleep 1994;17:S54-59.
8. Dauvilliers Y, Montplaisir J, Molinari N, Carlander B, Ondze B, Besset A, et al. Age at onset of narcolepsy in two large populations of patients in France and Quebec. Neurology 2001;57:2029-2033.
9. American Academy of Sleep Medicine. ICSD-2-International classification of sleep disorders (2nd ed). Diagnostic and coding manual. American Academy of Sleep Medicine; 2005.