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Case Report

# A Case of Narcolepsy in a Seven Year Old Boy

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## Abstract

Narcolepsy is a lifelong neurologic disorder of rapid eye movement (REM) sleep in which there are attacks of irresistible daytime sleepiness, cataplexy, hypnagogic hallucinations, sleep paralysis, and fragmented night sleep [1]. It can affect the school performance of children and adolescents causing their grades to drop. Sometimes they are mistakenly termed as lazy. In adults, narcolepsy can cause marital problems, social withdrawal, job termination and even accidents.

In this case report, we are reporting a case of seven year old with narcolepsy, this is the first case report for narcolepsy in 7 years old in Arab region.

***Keywords:*** *Narcolepsy; Excessive Daytime Sleepiness*

## Introduction

Narcolepsy is a lifelong neurologic disorder of rapid eye movement (REM) sleep in which there are attacks of irresistible daytime sleepiness, cataplexy, hypnagogic hallucinations, sleep paralysis, and fragmented night sleep [1]. It can affect the school performance of children and adolescents causing their grades to drop. Sometimes they are mistakenly termed as lazy. In adults, narcolepsy can cause marital problems, social withdrawal, job termination and even accidents.

## Case Presentation

Patient S, a 7 year old boy, referred to Pediatric Sleep Disorders Center at Prince Sultan Military City, for evaluation of excessive daytime sleepiness (EDS). He was well till he was 5-years old; he was noted to have prolonged sleeping time during the day and night. He would sleep for long periods of time in the car, airplane and suddenly while playing. He was noted to have abnormal movement like tongue deviation to the left while playing cellphone and watching TV. The patient sleeps an average of 10 hours per night in addition of taking a nap during the day for an average of 3 hours. He is also excessively sleepy and tired during the day.

The patient was seen initially at the age of five years in different hospitals with an impression of partial epilepsy and he was started on Phenobarbitone and Keppra for two years with no much improvement.

On physical examination, his height was 122 cm, weighed 29.05 kg with a BMI of 19.5 kg/m2 and his neck circumference is 34 cm. The patient looks well, conscious, oriented, with no signs of distress and was afebrile. No cyanosis or rashes in his skin, mild tongue protrusion was noted, and no nasal congestion. Upon chest examination, it is clear and equal in symmetry. His vital signs were within normal limits and with good oxygen saturation.

The patient underwent a series of blood tests to rule-out other problems. Overnight polysomnographic study was done which showed multiple REM periods (Figure 1) and prolonged Total Sleep Time (TST) with no evidence of Sleep-Disordered Breathing (SDB), nor periodic Leg Movements (PLMs) (Table 1 -4). MSLT was done the next day (Figure 2), which showed all naps had short duration for sleep latency and three sleep onset REM periods (SOREMPS) out of 4 naps, which was highly suggestive of narcolepsy. Hence the patient went through HLA-DQB1*0602 typing. Consultations were also done with the neurologist, endocrinologist, and geneticist, where secondary causes of narcolepsy were ruled out.

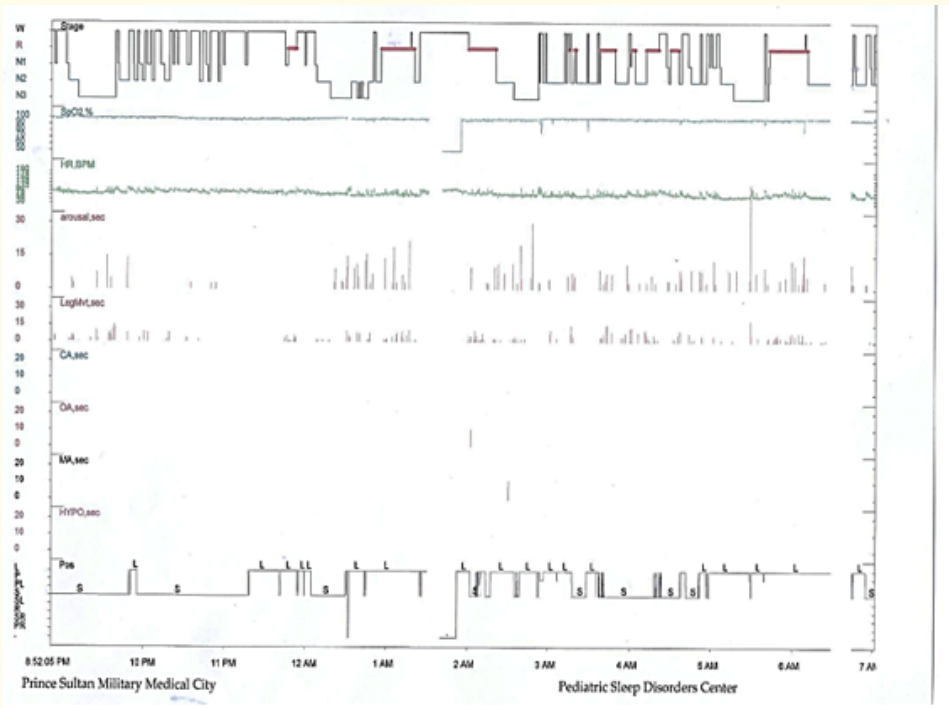


***Figure 1:*** *Sleep architecture during the PSG showing multiple REM periods (in red circle).*

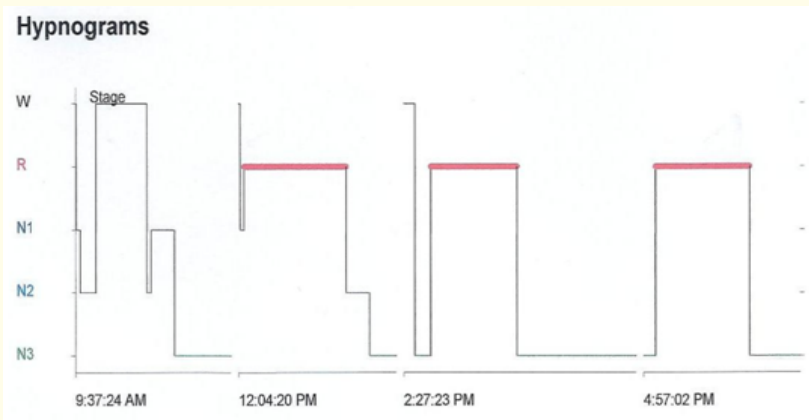


***Figure 2:*** *MSLT showing SOREMPs in naps 2,3 and 4. The Mean Sleep Latency is 25 seconds, and the Mean REM latency of 1.20 minutes which is strongly suggestive of narcolepsy.*

| Sleep Architecture | Normal Values | Results |
|---|---|---|
| Total Recording Time (TRT) | 6 - 8hrs or 360 - 480 minutes (may vary as per policy) | 632.4 minutes |
| Total Sleep Time (TST) | 9 - 10 hrs/24 hrs or 540 - 600 minutes/1440 minutes | 434.0 minutes |
| Sleep Efficiency | 85% - 90% or higher | 71.2% |
| Sleep onset | 10 - 20 minutes | 1.2 minutes |
| REM Latency (from Sleep Onset) | 90 - 120 minutes | 173 minutes |

***Table 1:*** *Sleep architecture.*

| Sleep Staging | Normal Values (% in Healthy Young Adults) | Duration | %TST |
|---|---|---|---|
| N1 | 2%-5% | 58.5 minutes | 13.5% |
| N2 | 45%-55% | 165.0 minutes | 38.0% |
| N3 | 13%-23% | 105.5 minutes | 24.3% |
| REM | 20%-25% | 105.0 minutes | 24.2% |

***Table 2:*** *Sleep staging.*

| Respiratory Data | Normal Values (Pediatric) | Results |
|---|---|---|
| Apneas | - | 1 |
| Hypopneas | - | 0 |
| AHI | < 1.5/hr | 0.1/hr |
| Mean $0_2$ Saturation | > 96% | 96% |
| Minimum $0_2$ Saturation | > 93% | 74% |

***Table 3:*** *PSG- respiratory data.*

| Limb Movements | Normal Values (Pediatric) | Count | Index (#/h) |
|---|---|---|---|
| Total Leg Movement | - | 108 | 10.7 |
| PLMS | < 1/hr TST | 33 | 3.3 |
| PLMS Arousals | - | 6 | 0.6 |

***Table 4:*** *PSG-limb movements.*

## Discussion

The correct prevalence of narcolepsy in children is not easy to establish. Yoss and Daly have reported 400 narcolepsy patients in the period from 1957 - 1960. The data has shown that 15 patients (4%) were below 15 years of age [2]. However, Challamel., et al. has shown that the onset of symptom started in 34% before the 15 years of age, 16% before the age of 10 and 4.5% before the age of 5 [3,4].

Yoss and Daly has shown that in preschool-aged narcolepsy is rare. As evidenced by 11.7% out of 85 subjects, were below 5 years of age [5]. Furthermore, narcolepsy is a very challenging task to confirm prior to the age of 4 to 5 years, because at this phase even unaffected kids have a habit of daytime naps and are unable to give precise history of cataplexy, hypnagogic hallucinations, or sleep paralysis.

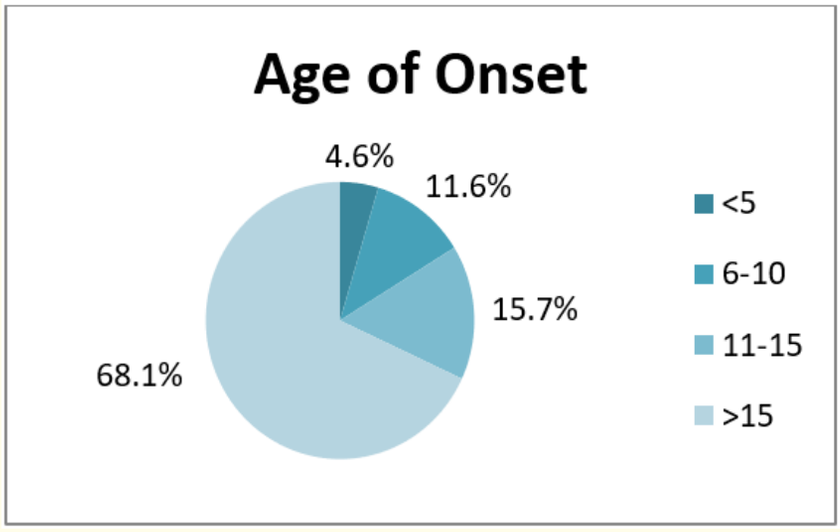


***Figure 3:*** *The age of onset of narcolepsy. (Adapted from Principles and Practices of Pediatric Sleep Medicine Stephen.*

In school-aged children, daytime sleepiness is the most common features of narcolepsy which consistent of foggy feeling from drowsiness and superimposed on this background are periods of more dramatic sleep attacks. However, cataplexy is the second most presenting features of narcolepsy [3], which is most specific features for the disease, and consists of sudden loss of muscle tone in the extensor muscles of the thighs, back, or neck in response to emotional changes.

Hypnagogic (sleep onset) hallucinations or hypnopompic (upon awakenings from sleep) as seen in 50-60% which manifests as either auditory or visual in nature. Sleep paralysis, however, which is a sudden momentary inability to move, is another presenting feature of narcolepsy. Sleep paralysis is drifting off to sleep or awakening from sleep. Young and colleagues have shown that sleep fragmentation secondary to periodic limb movements were found in 5 (63%) of 8 children with narcolepsy [6].

Narcolepsy was determined to result from genetic predisposition, abnormal neurotransmitter functioning and sensitivity, and abnormal immune modulation. Current data implicate certain human leukocyte antigen (HLA) subtypes and abnormalities in monoamine synaptic transmission, particularly in the pontine reticular activating system. The close HLA association of narcolepsy has led to the theory that narcolepsy is caused by an autoimmune destruction of hypocretin cells in susceptible individuals. The disorder is transmitted in an autosomal recessive fashion with full penetrance and is characterized mainly by cataplexy [7,8].

In literature, 85% to 95% found that there was a reduction in the number of hypocretin neurons in the hypothalamic region [9]. Nishino and coworkers, using the radioimmunoassay, found that the mean CSF level of hypocretin-1 in healthy controls was 280.3 + 33.0 pg/ml, and in neurologic controls was 260.5 + 37.1 pg/ml compared in those with narcolepsy, hypocretin-1 was either undetectable or below 100 pg/ml [10]. In the diagnostic sensitivity of low levels (less than 100 pg/Ml) the result was 84.2%. 32 out of 38 patients, were found HLA DQB1*0602 positive with low to absent levels of hypocretin. However, HLA-negative narcolepsy patients had a normal to high CSF hypocretin-1 levels.

Another study has shown that 92.3% of patients, who were both DQB1*0602-negative narcolepsy patients had normal to high CSF hypocretin-1 levels, while DBQ1*0602-negative patients without cataplexy had normal levels [11]. Kanbayashi and colleagues have shown that patients with narcolepsy without cataplexy and idiopathic hypersomnia patients had normal CSF hypocretin levels [12].

The diagnosis of narcolepsy is established on the basis of the history, combined with characteristic findings on the nocturnal polysomnogram and a multiple sleep latency test [13]. During the nocturnal polysomnogram, multiple parameters of physiologic activity are recorded simultaneously on a computerized sleep monitoring and analysis system [14]. Patients with narcolepsy may exhibit onset of REM sleep within 15mins of sleep onset. Gross sleep efficiency is generally higher (greater than 90%), but there may be fragmentation of sleep from an increased number of arousals and PLMS.A useful clue to narcolepsy in children is the presence of decreased nocturnal REM sleep latency [15].

Isolated cataplexy has been observed in Niemann-Pick type C disease, but these patients cannot be labeled as having secondary narcolepsy as they lack other clinical and polysomnographic features [16]. The most common disorder leading to excessive daytime sleepiness (EDS) in childhood is insufficient nocturnal sleep [17]. Superimposed with this may be elements of abnormal sleep hygiene or circadian rhythm disturbances such as the delayed sleep phase syndrome [18,19].

Narcolepsy needs a lifetime treatment, the overnight polysomnogram and MSLT results should be positive before making the diagnosis. Stimulants like methylphenidate (regular or extended-release formulations) or other preparations of amphetamine are used for excessive daytime sleepiness [20-23]. One or two planned naps per day for about 25 - 30 minutes had a great help to boost daytime alertness and improve psychomotor performance [24].

## Conclusion

In the last few years, there were different studies in order to figure out and treat narcolepsy. Abnormal genetic predisposition and neuropeptides are closely linked to the pathophysiology of narcolepsy. Narcolepsy in childhood is difficult to diagnose, thus needing prompt evaluation and further investigation for adequate treatment. Both pharmacological and non-pharmacological interventions should be applied to pediatrics.

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