

CASE: NARCOLEPSY

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CASE: A 30-year old woman is referred for evaluation of excessive daytime sleepiness for the past 10-12 years. She has become more concerned because of several “near miss” motor vehicle incidents. She is a college graduate with a degree in early childhood education. She is the director of a day care facility and works 8 AM to 6 PM five days per week. She has been in this job for the past 4 years. Her responsibilities include supervision of the teaching staff and a variety of administrative tasks. She also fills in for teachers during their breaks or vacations.

Sleepiness was first noted during her college years and was attributed to an erratic schedule and late night studying. For the past 8 years however despite a regular schedule, with a bedtime of 10:30 PM and wake time of 6:15 AM, the sleepiness persists. Although she wakes easily in the morning, by 9 AM she feels so sleepy that she takes a brief nap in the office (with the door shut). She sleeps for 15 minutes and wakes refreshed. This urge to nap recurs an additional two times daily. It is particularly difficult when she is filling in for a teacher and can't nap. She is usually aware of dreams during the naps.

At night she falls asleep quickly (5 minutes) and briefly awakens once or twice during the night. She describes frequent sleep paralysis when going to sleep. This also happens during many of her naps. She has often noticed a shadowy figure or image in the room when falling asleep at night. She has also experienced transient weakness, usually when laughing vigorously, several times over the past 8 years.

The patient walks to work, so her driving is limited, though she has had a couple close calls when driving around town on weekends. She drinks 4-5 cups of coffee throughout the day. She has 1-2 glasses of wine most evenings. She denies use of illicit drugs and is a non-smoker. She is otherwise healthy and is taking no medication other than oral contraceptives. She is aware of similar sleepiness in her younger brother (age 24). The physical examination is unremarkable.

- 1) Based on the history, what is the differential diagnosis?
- 2) What are the features that suggest narcolepsy?
- 3) What further diagnostic evaluation, if any, do you recommend?

The polysomnography revealed a total sleep time of 425 minutes with a 90% sleep efficiency. The REM sleep latency was 12 minutes. Sleep staging was normal and there were no other abnormalities. The MSLT revealed a mean sleep latency of 3.4 minutes and there were 3 sleep-onset REM periods.

- 4) Are the sleep laboratory findings helpful?
- 5) How would you treat this patient?
- 6) What would you tell this patient about the general prognosis and treatment expectations?

Treatment for sleepiness was initiated with methylphenidate and the patient was encouraged to maintain a regular schedule with adequate time in bed each night. Since the auxiliary symptoms were rather frequent and quite troubling, she was also prescribed fluoxetine. Her sleepiness improved significantly with methylphenidate, though she still elected to minimize driving. She has had few episodes of cataplexy when she ran out of fluoxetine.

TEACHING POINTS:

- 1) Sleepiness in a young patient, who does not have evidence of insufficient sleep or schedule disturbance, is suggestive of narcolepsy. The usual onset of sleepiness in narcolepsy is in the late teens or early twenties, consistent with this patient's presentation. Her current sleep schedule should allow for adequate sleep otherwise, and the schedule appears to be stable across the week. The presence of sleep paralysis, hypnagogic imagery and cataplexy are all consistent with the diagnosis of narcolepsy. Cataplexy is thought to be pathognomonic for narcolepsy. Sleepiness and the REM sleep auxiliary features are referred to as the narcolepsy tetrad. Other considerations at the outset of the history might have included insufficient sleep, schedule disturbance, idiopathic hypersomnolence and underlying sleep disruptors (to account for the reported nocturnal awakenings).
- 2) While narcolepsy is a clinical diagnosis, polysomnography (PSG) and multiple sleep latency test (MSLT) are recommended to exclude additional underlying sleep disruptors (by PSG), and objectify the sleepiness complaint and identify sleep onset REM periods (SOREMs) that are characteristic of the disorder (by MSLT).
- 3) The test results are supportive of the clinical diagnosis of narcolepsy. There is no evidence of sleep rebound, nor other sleep disruptors. There is a SOREM on the PSG and very severe sleepiness with 3 SOREMs on the MSLT.
- 4) Patients with narcolepsy should be cautioned about maintaining adequate sleep time (as in any other patient). They will generally however require medication to offset the sleepiness. Agents commonly used include methylphenidate, modafinil and amphetamines. In a patient with troublesome REM sleep auxiliary features (sleep paralysis, hypnagogic imagery and or cataplexy) additional treatment with a REM sleep suppressing medication is warranted. These typically include the SSRIs or TCAs.
- 5) Understanding narcolepsy as a lifelong condition, and the role of medication in controlling symptoms are important in the satisfactory management of the disorder. The familial risks should be addressed, and driving (or other high risk activities) issues should be discussed as well.