

CMEO BriefCase

Diagnostic Tools: A Process of Exclusion

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Learning Objective

Integrate appropriate assessments and tools to facilitate early diagnosis of IH

Patient Case: Maria



- 26-year-old Hispanic female presents with complaints of “being tired”, “brain fog”, despite 11-12 hours of sleep per day. Complaints have been persistent for many years.
- “It’s really hard to wake up for alarms, and when I do, I don’t feel awake for a long time. I have trouble functioning, and often can’t think or remember things”
- Patient was recently let go from her job due to consistently underperforming.
- Symptoms emerged ten years prior, diagnosis of depression given three years ago, treatment initiated at this time with some improvement for depression, but not sleep symptoms.
- Labs (2 weeks prior): CBC, CMP, Iron panel, Vit B12, TSH – WNL
- Current findings: BP = 130/82, BMI = 29, PHQ-9 = 10
- Medications: Fluoxetine 60mg once daily

BMI = body mass index; BP = blood pressure; CBC = complete blood count; CMP = comprehensive metabolic panel; PHQ-9 = Patient Health Questionnaire; TSH = thyroid stimulating hormone; WNL = within normal limits

Audience Response



What characteristic associated with Maria's case indicates idiopathic hypersomnia specifically as a potential diagnosis?

- A. Long sleep time
- B. Depression
- C. Onset of symptoms in teenage years
- D. Cognitive impairment
- E. I don't know

Presentation of Idiopathic Hypersomnia

Signs/Symptoms

"I am always tired"



Sleep > 10 hours of sleep



Sleep is not refreshing



Sleep inertia

(sleep drunkenness)



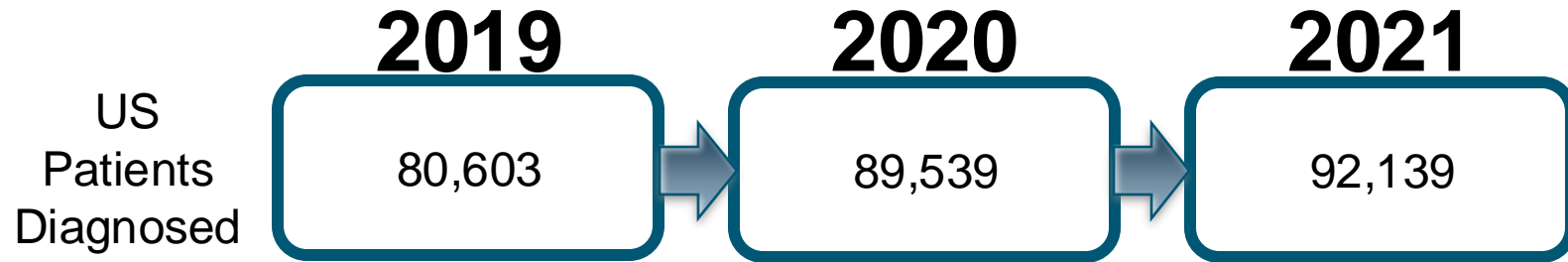
Brain fog
Cognitive impairment



Often using
non-prescription
measures to stay awake



Prevalence and Burden



*More prevalent in women, higher prevalence of sleep apnea, mood disorders, headache, migraine, and cardiovascular disease

ARISE study

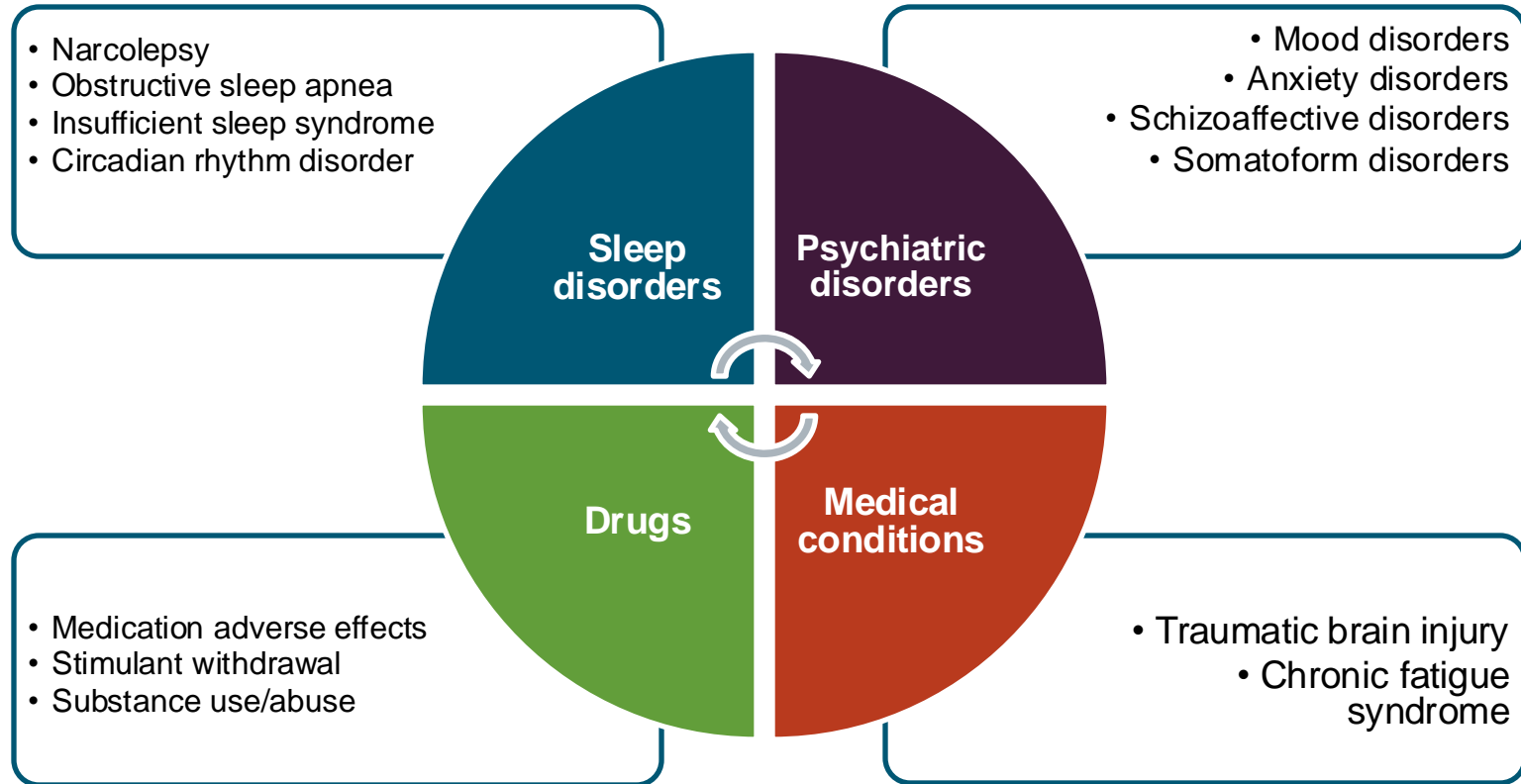
Patients with long sleep time vs. without long sleep time experience:

- Lower QoL scores for social life and stigma
- More severe cognitive complaints
- More cases of severe depression
- Worse presenteeism and activity impairment

QoL = quality of life

Lillaney P et al. *SLEEP*. 2023;46(Supplement_1):A373. Saad R et al. *SLEEP*. 2023;46(Supplement_1):A369. Schneider L et al. *Neurology*. 2023;100(17 Supplement 2)

Differential Diagnosis of IH



Audience Response



Which are diagnostic criteria designated by the ICSD-3 that are exclusive to IH vs. narcolepsy?

- A. Daily periods of irrepressible need to sleep or daytime lapses into sleep, present for at least 3 months
- B. No cataplexy is present
- C. Fewer than two SOREMPs on MSLT
(or fewer than one if nocturnal REM latency was ≤ 15 min)
- D. I don't know

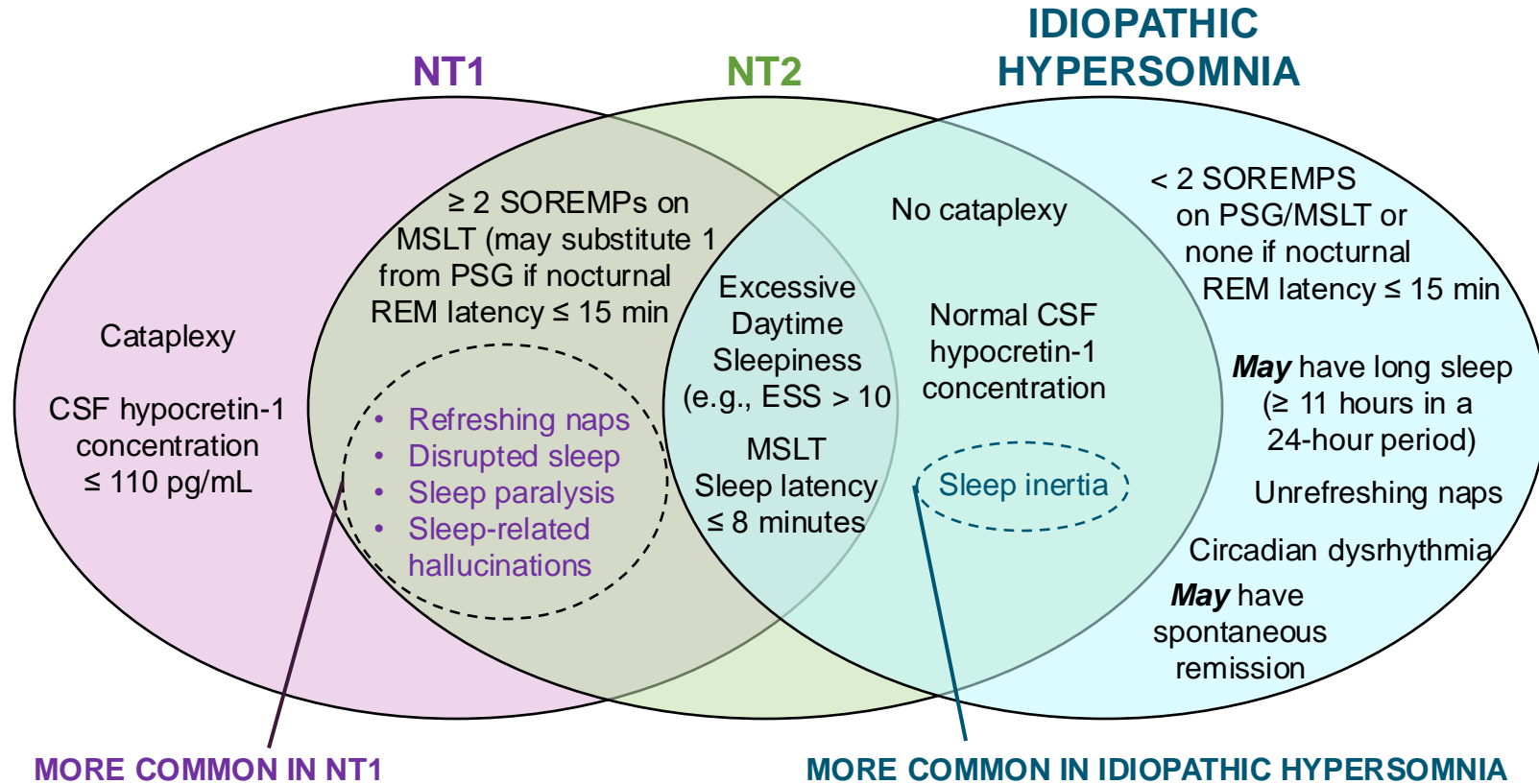
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Differentiation Between Narcolepsy and IH



CSF = cerebrospinal fluid; ESS = Epworth Sleepiness Scale; MSLT = Multiple Sleep Latency Test; PSG = polysomnography; REM = rapid eye movement; SOREMPs = sleep onset REM periods
 Dauvilliers Y et al. *Sleep Med Rev.* 2022;66:101709.

IH Diagnosis: ICSD-3 Criteria/Limitations

Current Approach

- A. **Daily periods of irrepressible need to sleep or daytime lapses into sleep, present for at least 3 months** → Same item A for NT1/2!
Unidimensional aspect? No hypersomnia?
- B. Fewer than two SOREMPs on MSLT (or fewer than one if nocturnal REM latency was ≤ 15 min) → Number of SOREMPs variable between tests
- C. No cataplexy
- D. At least one of the following: → Wrist actigraphy: Not objective sleep assessment
1. Mean sleep latency ≤ 8 min on MSLT
 2. **Total 24-h sleep time ≥ 660 min on 24-h PSG or wrist actigraphy (averaged over ≥ 7 d)** → **Which causes? How to be ruled out?**
- E. Insufficient sleep syndrome is ruled out
Sleep restriction, mild AHI, mild PLMS, Low sleep efficiency, low TST on PSG?
- F. The hypersomnolence and/or MSLT findings are not better explained by other causes
NT2: Diagnosis because of MSLT
Depressive symptoms: Consequences?
Obesity, CNS drugs intake

AHI = apnea-hypopnea index; CNS = central nervous system;
PLMS = periodic limb movements of sleep; TST = total sleep time
Sateia MJ. *Chest*. 2014;146(5):1387-1394.

Idiopathic Hypersomnia Severity Scale (IHSS)

NIGHTTIME:

1. What is your ideal duration of night-time sleep ?
2. Do you feel that you have not had enough sleep?

AWAKENING:

3. Is it extremely difficult, to wake in the morning?
4. How long does it take you to function properly after you get up?
5. After waking up, do you ever do or say irrational things, and/or are you very clumsy?

NAPS:

6. Do you ever take a nap?
7. What is the ideal length of your naps ?

8. How do you feel after a nap?

9. During the day, do you ever struggle to stay awake?

BURDEN:

10. Does your hypersomnolence has an impact on your general health?

11. Is your hypersomnolence a problem in terms of your proper intellectual functioning?

12. Does your hypersomnolence affect your mood?

13. Does your hypersomnolence prevents you from carrying out daily tasks properly?

14. Does your hypersomnolence affect your driving a car?

0 → 12
MILD

13 → 25
MODERATE

26 → 38
SEVERE

39 → 50
VERY SEVERE

**Cut off to discriminate
IH and controls: 22**

Sensitivity: 91.1%

Specificity: 94.5%

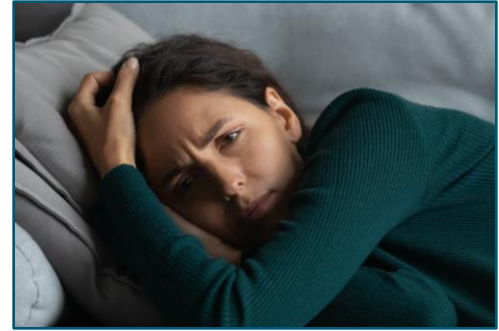
*For the IHSS score, the minimal clinically important difference (MCID) is 4 points

IH Diagnostic Challenges



- IH vs. narcolepsy
 - Daily periods of irrepressible need to sleep or daytime lapses into sleep present for at least 3 months → same in IH, NT1, and NT2 (ICSD-3)
- SOREMPS are variable between tests
- Diagnostic tools
 - PSG is rarely performed to measure maximal sleep amount
 - MSLT assesses daytime sleep propensity, not sleep inertia / long sleep time
 - Challenges in how to assess sleep inertia
 - Wrist actigraphy accuracy may vary by degree of sleep efficiency; most accurate when sleep efficiency is high
 - Sleep logs may provide discrepant reports compared to actigraphy, patients may not reliably complete sleep logs
- Few studies recorded patients with 24-hr protocol recording
 - With different protocols, it is not always standardized
 - Limitations in defining a pathological threshold for IH

Patient Case: Revisiting Maria



- 26-year-old Hispanic female presents with persistent fatigue, cognitive impairment, excessive daytime sleepiness despite > 10 hrs of sleep per day.
- What would we rule out?
- 24-hour sleep study conducted, findings:
 - MSLT: 1 sleep-onset REM period
 - Sleep latency: 6 minutes
 - Total sleep time: 702 minutes
- Other pertinent findings?

Diagnosis?

SMART Goals



- Differentiate idiopathic hypersomnia from other medical disorders, psychiatric disorders, sleep disorders, and medication/substance use.
- Distinguish between the subtleties of idiopathic hypersomnia, and narcolepsy types 1 and 2.
- Incorporate the ICSD-3 guidelines into practice when diagnosing patients suspected of having idiopathic hypersomnia.
- Recognize diagnostic tools utilized for idiopathic hypersomnia and the limitations of these tools.

CMEO  **BriefCase** **2**

Recognizing Idiopathic Hypersomnia: The Patient Journey to Diagnosis

CMEO  **BriefCase** **3**

Choosing Treatment for Idiopathic Hypersomnia: Matching Needs to Therapy

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<https://www.cmeoutfitters.com/practice/sleep-disorders-hub/>

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