

CMEO BriefCase

Recognizing IH: The Patient Journey to Diagnosis

*Supported by an independent medical
educational grant from Jazz Pharmaceuticals.*

Michael J. Thorpy, MD

Professor of Neurology

Albert Einstein College of Medicine

Director, Sleep-Wake Disorders Center, Department of Neurology

Montefiore Medical Center, Bronx, NY

President, New York State Society of Sleep Medicine

Past President, Sleep Section of the Academy of Neurology

Bronx, NY

Paul Doghramji, MD, FAAFP

Senior Family Physician

Collegeville Family Practice

Medical Director, Health Services

Ursinus College

Collegeville, PA

Learning Objective

Screen for idiopathic hypersomnia based on clinical presentation of patient or family/caregiver description of function and changes in quality of life.

Patient Case: Naomi

- 22-year-old Hispanic female presents with complaints of being “tired all of the time” despite >10 hours of sleep per day
- *“I have low energy and ability to focus and stay organized. Naps do not refresh me. I haven’t been doing well in college because of all this. I am often depressed because of everything going on.”*
- Pediatrician consult at age 16; fatigue was attributed to long hours spent in extracurricular activities, studying for classes
- PMH: depression
- Findings: BP 122/80, PHQ-9 = 14, BMI = 29
- Medications: birth control



Audience Response



What about Naomi's presentation is specific to idiopathic hypersomnia?

- A. Comorbid depression
- B. Inability to focus
- C. Excessive daytime sleepiness
- D. Unrefreshing naps
- E. I don't know

Audience Response



What about Naomi's presentation is specific to idiopathic hypersomnia?

- A. Comorbid depression
- B. Inability to focus
- C. Excessive daytime sleepiness
- D. **Unrefreshing naps**
- E. I don't know

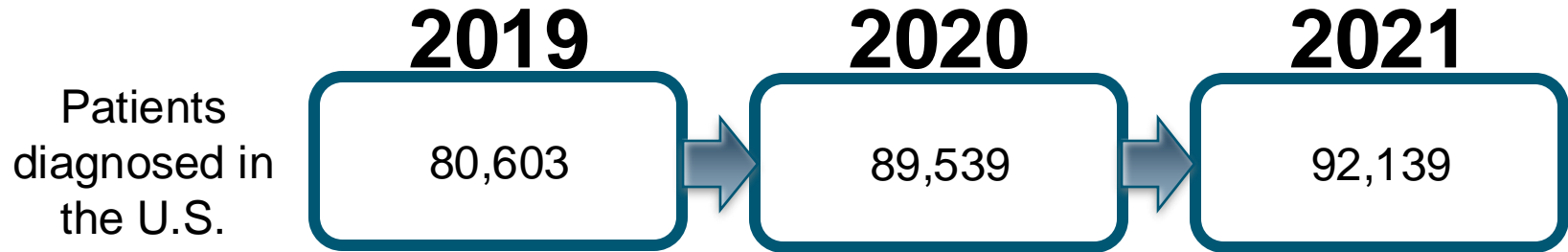
Patient Presentation Explained by Patients

- *“I can’t turn my sleep switch off”* – Shelly → EDS
- *“I don’t ever remember a time when I woke up feeling refreshed”* – Teresa → EDS after >10 hours of daily sleep
- *“The biggest loss is true human exchange which requires remembering, processing information...finding my words quickly and succinctly, following a joke... or engaging in an exchange of activity”* – Robyn → Brain fog, cognitive impairment
- *“This is what idiopathic hypersomnia feels like for me, a general anesthetic”* – Denise → Sleep inertia

EDS = excessive daytime sleepiness

Hypersomnolence Australia. Hypersomnolence Australia Website. 2023. <https://www.hypersomnolenceaustralia.org.au/single-post/2018/09/03/thoughts-and-stories-by-people-living-with-idiopathic-hypersomnia>

Prevalence and Burden



Higher prevalence in women

Greater prevalence of:

- Sleep Apnea
- Headache Migraine
- Mood Disorders
- Cardiovascular Disease

Hypersomnia in Children



Characteristic	CH (n = 33)	Controls (n = 33)
Narcolepsy with cataplexy	7	
Narcolepsy without cataplexy	11	
IH diagnosis	15	
Ratio, male/female	16:17	16:17
Age, mean (SD; range), years	12.93 (2.76; 8.03–16.84)	12.74 (2.75; 8.38–17.81)
Race/Ethnicity, White/African American/Hispanic	16:15:2	18:15:0
Household income, mean (SD), USD	\$33,550.91 (\$11,982.76)	\$36,848.97 (\$13,904.82)

Pediatric QoL Study Results

Outcome	Narcolepsy/IH	Control	P-value
Sleepiness (ESS)	12.3	6.9	< .001
BMI	26.2	22.2	.03
Injury history	39%	15%	.03
Physical activities	30%	56%	.04
Extracurricular activities	25%	68%	.001

Outcome	Narcolepsy/IH	Control	P-value
Academic grades < C	25%	12.5%	.04
Physical functioning	74.5	85.5	.001
Social functioning	75.8	87.8	.01
School functioning	65.9	78.1	.007
Overall QoL	73.4	83	.001

CH = central hypersomnia; ESS = Epworth Sleepiness Scale; IH = idiopathic hypersomnia; QoL = quality of life; USD = United States dollars

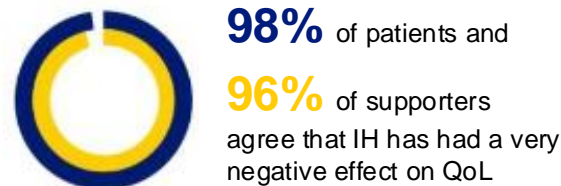
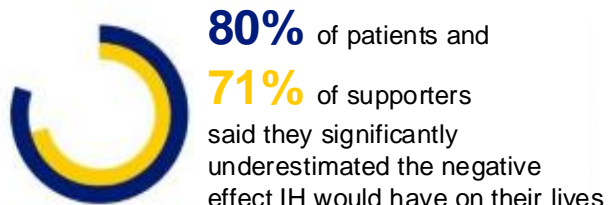
Avis KT, et al. *J Clin Sleep Med*. 2015;11(11):1281-1288.

Parents and Family Members Struggle Too

Pediatric QoL Study Results: Parents

Outcome	Narcolepsy/IH Parent	Control Parent	P-value
Physical functioning	62.5	80.6	< .001
Emotional functioning	58.9	70.8	.007
Social functioning	64.8	81.2	.002
School functioning	49.8	77.7	< .001
Overall QoL	59.5	78	< .001

The results from the “I Have IH” patient and supporter surveys revealed the far-reaching impact of IH:



Struggle into Adulthood



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

“I Have IH”
Patient Survey
Results



Audience Response



For roughly 1 out of every 5 patients diagnosed with IH, how long does it take to receive their diagnosis after seeking care for their symptoms?

- A. Within one year
- B. Up to five years
- C. Up to ten years
- D. More than ten years
- E. I don't know

Audience Response



For roughly 1 out of every 5 patients diagnosed with IH, how long does it take to receive their diagnosis after seeking care for their symptoms?

- A. Within one year
- B. Up to five years
- C. Up to ten years
- D. **More than ten years**
- E. I don't know

Patients Wait Years for Diagnosis



- Results from the patient and supporter survey showed how long it took patients to receive an IH diagnosis after seeking medical care for their symptoms:

- The results from the patient and supporter surveys revealed that:



31%

of patients said they received their IH diagnosis with **1 year**

37%

said it took up to **5 years**

32%

said it took anywhere from **5** to more than **10 years**

19%
of those with IH had to wait more than **10 years** after seeking medical care to get their diagnosis



Nearly

TWO-THIRDS

of patients (**61%**) and supporters (**63%**) strongly or somewhat agreed that they or the person they care for were

MISDIAGNOSED

with other medical conditions **before being diagnosed with IH**



Screening Tools: Epworth Sleepiness Scale

Epworth Sleepiness Scale

Name: _____ Today's date: _____

Your age (Yrs): _____ Your sex (Male = M, Female = F): _____

How likely are you to doze off or fall asleep in the following situations, in contrast to feeling just tired?

This refers to your usual way of life in recent times.

Even if you haven't done some of these things recently try to work out how they would have affected you.

Use the following scale to choose the **most appropriate number** for each situation:

0 = would **never** doze
1 = **slight chance** of dozing
2 = **moderate chance** of dozing
3 = **high chance** of dozing

It is important that you answer each question as best you can.

Situation	Chance of Dozing (0-3)
Sitting and reading _____	
Watching TV _____	
Sitting, inactive in a public place (e.g. a theatre or a meeting) _____	
As a passenger in a car for an hour without a break _____	
Lying down to rest in the afternoon when circumstances permit _____	
Sitting and talking to someone _____	
Sitting quietly after a lunch without alcohol _____	
In a car, while stopped for a few minutes in the traffic _____	

- 0-5: Lower normal daytime sleepiness
- 6-10: Higher normal daytime sleepiness
- 11-12: Mild excessive daytime sleepiness
- 13-15: Moderate excessive daytime sleepiness
- 16-24: Severe excessive daytime sleepiness

Screening Tools: (FOSQ-10/FOSQ-30)

- Scoring measures functionality of patient in relation to their sleepiness
- Measure of HRQoL
- Consists of shorter 10 question survey (FOSQ-10), or longer 30 question survey (FOSQ-30)

Functional Outcomes of Sleep Questionnaire (FOSQ) (reduced)					
Please mark "X" as appropriate:		1	2	3	4
		Yes, extreme	Yes, moderate	Yes, a little	No
1	Do you have difficulty concentrating on the things you do because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
2	Do you generally have difficulty remembering things because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
3	Do you have difficulty finishing a meal because you become sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4	Do you have difficulty working on a hobby (For Example: Sewing, Collecting, Gardening, etc.) because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
5	Do you have difficulty doing work around the house (For Example: Cleaning House, Doing Laundry, Taking Out the Trash, Repair Work, etc.) because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
6	Do you have difficulty operating a motor vehicle for short distances (less than 100 miles) because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
7	Do you have difficulty operating a motor vehicle for long distances (greater than 100 miles) because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
8	Do you have difficulty getting things done because you are too sleepy or tired to drive or take public transportation?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
9	Do you have difficulty taking care of financial affairs and doing paperwork (For Example: Writing Checks, Paying Bills, Keeping Financial Records, Filling Out Tax Forms, etc.) because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
10	Do you have difficulty performing employed or volunteer work because you are sleepy or tired?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
FOSQ - 10 Score					

FOSQ = Functional Outcomes of Sleep Questionnaire; HRQoL = health-related quality of life

Omachi TA. *Arthritis Care Res (Hoboken)*. 2011;63 Suppl 11(0 11):S287-S296.

Image adapted from: https://batonrougeclinic.com/wp-content/uploads/2023/02/Functional-Outcomes-of-Sleep-Questionnaire-FOSQ_Update-02-01-2023.pdf

Idiopathic Hypersomnia Severity Scale

NIGHTTIME:

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

AWAKENING:

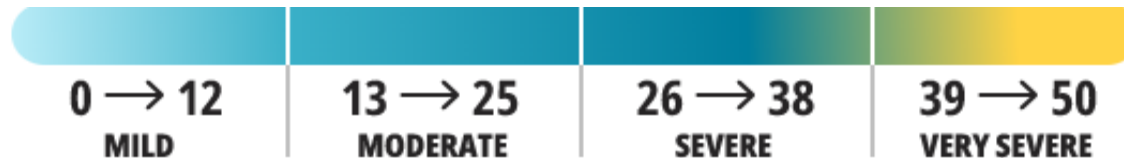
3. Is it extremely difficult, to wake in the morning?
4. How long does it take you to function properly after you get up?
5. After waking up, do you ever do or say irrational things, and/or are you very clumsy?

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?



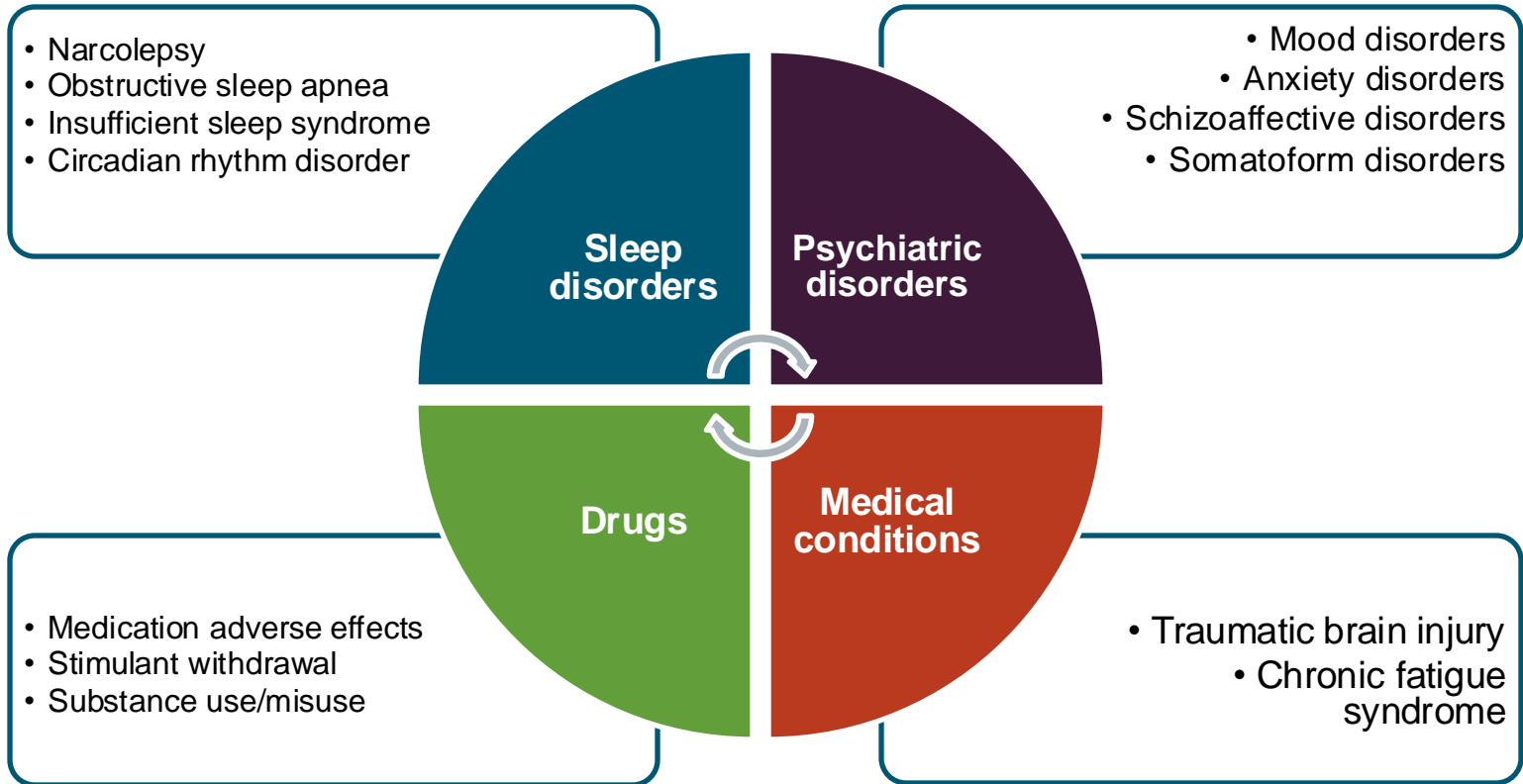
**Cut off to discriminate
IH and controls: 22**

Sensitivity: 91.1%

Specificity: 94.5%

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

Differential Diagnosis of IH



Patient Case: Naomi

- 22-year-old Hispanic female presents with complaints of being “tired all of the time” despite >10 hours of sleep per day



What about her presentation causes us to suspect a sleep disorder?

What specifics make us consider IH as a possibility?

What are our next steps?

SMART Goals



- Practice identification of patient symptoms, QoL factors, and family reports indicative of IH in your regular patient visits
- Improve screening practices for IH based on population-specific burdens in pediatric and adult patients
- Utilize objective screening tools in clinical practice that effectively detect sleep disorders and streamline the referral process
- Develop appropriate plan of action for IH suspected patients based on differential diagnosis and clinical findings

Series on Idiopathic Hypersomnia

CMEO
BriefCase **2**



*Diagnostic Tools: A
Process of Exclusion*

CMEO
BriefCase **3**



*Choosing Treatment:
Matching Needs to
Therapy*

www.CMEOutfitters.com/sleep-disorders-hub/

Sleep Disorders Education Hub

A robust hub of patient education and resources for your patients to learn more about idiopathic hypersomnia

cmeoutfitters.com/practice/sleep-disorders-hub/

To Receive Credit

To receive CME/CE credit for this activity, participants must complete the post-test and evaluation online.

Participants will be able to download and print their certificate immediately upon completion.