

# Diagnosis Journey, Symptoms, and Burden of Idiopathic Hypersomnia: Patient Perspectives From Qualitative Interviews

Michael J. Doane,<sup>1</sup> Meaghan O'Connor,<sup>2</sup> Miranda Lauher-Charest,<sup>2</sup> Laura Tesler Waldman,<sup>2</sup> Wilbur P. Williams, III<sup>1</sup>

<sup>1</sup>Alkermes, Inc., Waltham, MA, USA; <sup>2</sup>IQVIA Quality Metric Inc., Johnston, RI, USA

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## INTRODUCTION

- Idiopathic hypersomnia (IH) is a rare, chronic neurological disorder characterized by excessive daytime sleepiness (EDS), unrefreshing sleep, sleep inertia, cognitive impairments, and fatigue, among other symptoms<sup>1,2</sup>
- The symptom overlap with other disorders (eg, sleep disorders, mental health disorders) can further burden patients, as they often experience years of doctor visits, testing, and misdiagnoses before receiving an accurate diagnosis<sup>1,3,4</sup>
- Patients with IH may experience a diminished quality of life, with negative impacts on physical functioning, energy levels, social interactions, mental health, and personal relationships<sup>3,5</sup>
- To date, most research on the impact of IH has been collected and reported on using quantitative methods<sup>4</sup>; the lack of studies examining the patient experience using qualitative methods has resulted in a gap in the literature on this uncommon and often debilitating condition

## OBJECTIVE

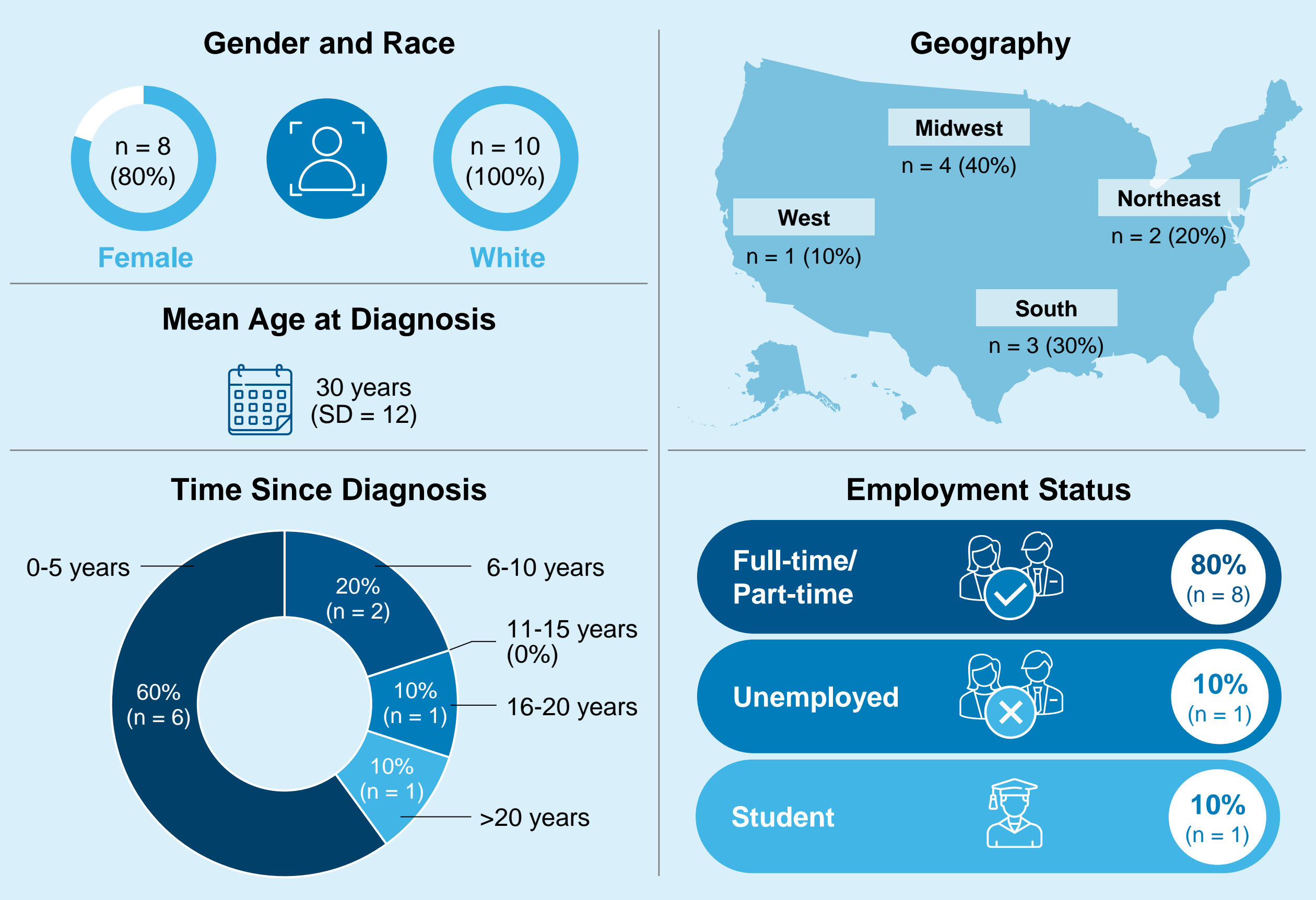
- The objective of this study was to use qualitative methods to characterize the lived experiences of people with IH, including their diagnostic journey, symptoms, and the impact of IH on their health-related quality of life

## RESULTS

### PARTICIPANT CHARACTERISTICS

- A total of 10 US-based adults with IH participated in this study (mean [SD] age: 37 [13] years) (**Figure 1**)
  - All participants (n = 10, 100%) self-identified as White, and the majority (n = 8, 80%) were women
  - Most participants (n = 6, 60%) self-reported being diagnosed within the last 5 years, and 80% (n = 8) of participants were employed either full-time or part-time
  - Most participants (n = 7, 70%) provided a confirmation of their IH diagnosis, while 3 (30%) indicated a physician has diagnosed them with IH

**FIGURE 1: Participant Demographics and Clinical Characteristics**



### JOURNEY TO DIAGNOSIS

- Half of the participants reported waiting ≥10 years from symptom onset to receive a diagnosis of IH (n = 5, 50%; **Figure 2**)
- Participants reported that symptom onset occurred most often in either adolescence (n = 4, 40%) or emerging adulthood (n = 3, 30%)
- Initial symptoms of IH commonly reported by participants included EDS (n = 7, 70%), fatigue (n = 3, 30%), and oversleeping (n = 3, 30%)
- During the diagnostic journey, participants sought care from various healthcare professionals, including sleep specialists (n = 5, 50%), psychiatrists (n = 4, 40%), and primary care physicians (n = 6, 60%)
- 70% of participants (n = 7) recalled that, prior to their IH diagnosis, they were misdiagnosed with conditions including depression (n = 3, 30%), sleep apnea (n = 2, 20%), chronic fatigue syndrome (n = 2, 20%), and upper airway resistance syndrome (n = 2, 20%)

**FIGURE 2: Time From Symptom Onset to IH Diagnosis**



IH = idiopathic hypersomnia.

### References

- Dauvilliers Y, et al. *Sleep Med Rev.* 2022;66:101709. 2. Trotti LM. *Sleep Med Clin.* 2017;12(3):331-344. 3. Whalen M, et al. *Sleep.* 2022;45(suppl 1):A175-A176. 4. Boulanger T, et al. *Sleep Adv.* 2024;5(1):zpa059. 5. Miglis MG, et al. *J Clin Sleep Med.* 2020;16(5):749-756.

## METHODS

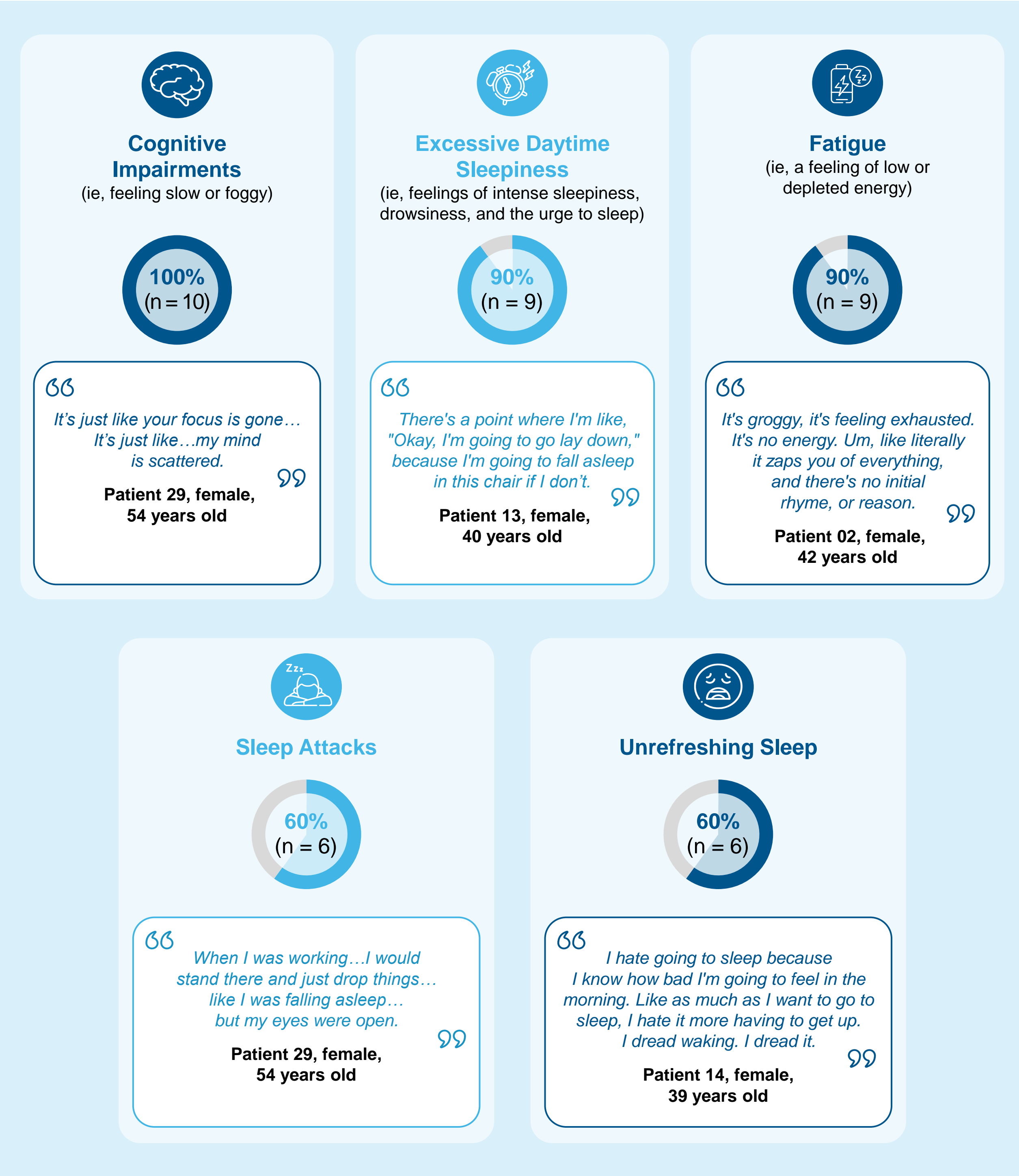
### STUDY DESIGN

- This was a qualitative, cross-sectional, observational study
  - Trained qualitative researchers conducted 60-minute concept elicitation interviews with adults with IH
  - Interviewers followed a semi-structured interview guide designed specifically for this study
- Study participants were recruited using purposive and snowball sampling, non-probability sampling methods commonly used in qualitative research; recruitment messaging was shared with potential participants through existing panels of patients, physician referrals, and social media outlets
  - Snowball sampling was included as a sampling method given the rare nature of IH
- Sample size determinations were based on the number of interviews anticipated to reach concept saturation
  - Saturation was assessed by dividing participant interviews across 2 sets of 5 interviews and comparing emerging concepts across sets
  - The saturation analysis suggests saturation of concepts was reached with 10 interviews and additional interviews would not have yielded more information
- During screening, participants self-reported the month and year of their IH diagnosis
- Transcribed participant interviews were coded and analyzed using inductive and deductive approaches (addressing pre-identified concepts while also capturing new concepts that emerged from the data)
- This study was approved by an institutional review board and all participants provided informed consent

### FREQUENCY OF SYMPTOMS

- The most frequently reported symptom was cognitive impairments, described as feeling “slow” or “foggy” (n = 10, 100%) (**Figure 3**)
  - Participants described experiencing lapses in memory (n = 4, 40%), difficulties with focus and attention (n = 4, 40%), trouble thinking things through (n = 6, 60%) and processing information (n = 9, 90%), and difficulty concentrating (n = 3, 30%)
- Participants also reported experiencing EDS (ie, profound sleepiness, drowsiness, and the desire to sleep; n = 9, 90%), and fatigue, described as a feeling of low or depleted energy (n = 9, 90%)
- Patients with IH described taking different medications to manage their symptoms, most commonly amphetamines (n = 7, 70%), armodafinil (n = 3, 30%), and modafinil (n = 2, 20%)

**FIGURE 3: Frequency of Symptoms Experienced by at Least Half of Participants With IH at Any Point in Their Patient Journey**

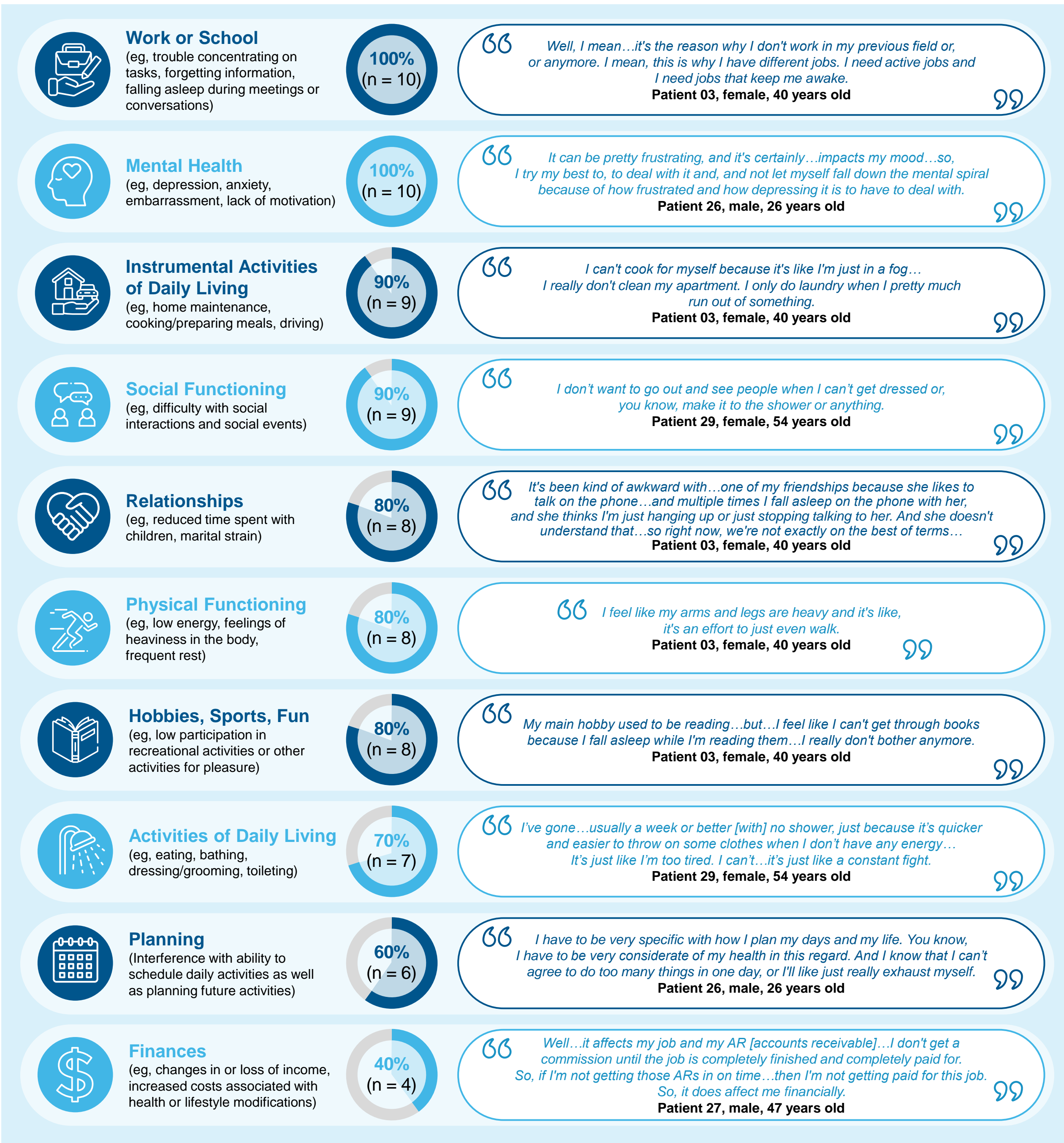


IH = idiopathic hypersomnia.

### PATIENT PERSPECTIVE — IMPACT ON QUALITY OF LIFE

- All, or nearly all, participants reported that IH negatively impacted their work and school activities (n = 10, 100%), mental health (n = 10, 100%), instrumental activities of daily living (n = 9, 90%), and social functioning (n = 9, 90%) (**Figure 4**)
- Most participants also described negative impacts on their relationships with family, friends, and intimate partners (n = 8, 80%), physical functioning (n = 8, 80%), their ability to engage in hobbies and/or recreational activities (n = 8, 80%), and activities of daily living (n = 7, 70%) as a result of their IH (**Figure 4**)

**FIGURE 4: Proportion of Participants With IH Reporting Impacts on Their Quality of Life**



IH = idiopathic hypersomnia.

## STUDY LIMITATIONS

- Recruiting study participants was challenging due to the rare nature of IH
  - Despite this limitation, a saturation analysis suggested saturation of concepts was reached and additional interviews with participants with IH would likely not have provided additional insights
- Participants included in the study were mostly female, all White, and the mean age was <40 years
  - It is unclear if results of the study would have been different with a broader participant population regarding gender, race/ethnicity, and age
- Confirmation of diagnosis of IH was encouraged but not required

## CONCLUSIONS

- This study provides a detailed account of the personal experiences of patients with IH encompassing the challenges patients encounter in their diagnostic journey and the most common symptoms they experience; it also provides an understanding into the depth and breadth of the impact IH has on their daily lives
- These results contribute to bridging gaps in the existing literature by offering meaningful insights through qualitative measures that authentically capture the lived experience of patients with IH

### Disclosures

MJD and WPW III are employees and shareholders of Alkermes, Inc. MO'C, ML-C, and LTW are or were employees of IQVIA Quality Metric, Inc. at the time this research was conducted.

