Diagnosis and Symptoms of Narcolepsy From the Patient Perspective: Results From In-Depth Qualitative Interviews

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INTRODUCTION

- Narcolepsy is a rare, chronic neurological condition characterized by excessive daytime sleepiness (EDS) and sleep-onset rapid eye movement (REM) periods[1,2]
- Narcolepsy is categorized into narcolepsy type 1 (NT1) and narcolepsy type 2 (NT2)[2]
  - Both types are characterized by EDS (including sleep attacks), sleep inertia, and sleep paralysis and/or hallucinations[2]
  - In addition, NT1 features cataplexy, which is a sudden, spontaneous, and temporary loss of muscle control triggered by strong emotional stimuli (e.g., fear, anger, laughter, or stress)[2,3]
- Symptoms of narcolepsy often start in adolescence or early adulthood, but can occur at any time[4]
- Lack of symptom recognition by clinicians may lead to misdiagnosis, potentially delaying effective treatment and further exacerbating disease burden[5]
- Previous studies that identified symptoms of narcolepsy have largely relied on quantitative methods such as surveys, which may provide limited insight into the patient experience compared to qualitative methods

OBJECTIVE

- The objective of this study was to use qualitative research methods to characterize the patient experience of adults living with NT1 and NT2
- The data presented herein reflect the diagnosis journey and symptom burden of narcolepsy

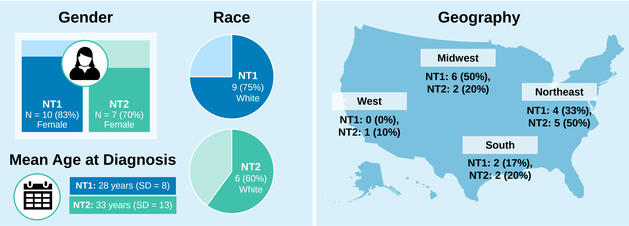
METHODS

- This was a qualitative, cross-sectional, observational study
  - Using a semi-structured interview guide, interviewers asked open-ended questions to gather insights into people's experiences with NT1 or NT2
- Adult participants with NT1 or NT2 were recruited from panels of patients, physician referrals, and social media outlets
  - Snowball sampling, whereby participants could refer other participants to this study, was also used
- Interview transcripts were coded and thematically analyzed using inductive and deductive approaches
- This study was approved by an Institutional Review Board and all participants provided informed consent

RESULTS

PARTICIPANT DEMOGRAPHICS AND DISPOSITION

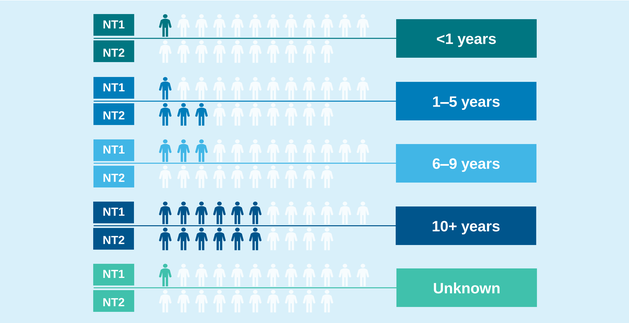
Figure 1. Demographics and Clinical Characteristics Among Participants



- A total of 22 adults with narcolepsy participated in this study (NT1 = 12; NT2 = 10)
- Overall, more than 70% of participants were female, and more than 60% of participants were White
- 83% of participants with NT1 and 50% of participants with NT2 were diagnosed with narcolepsy within the last 10 years

JOURNEY TO DIAGNOSIS

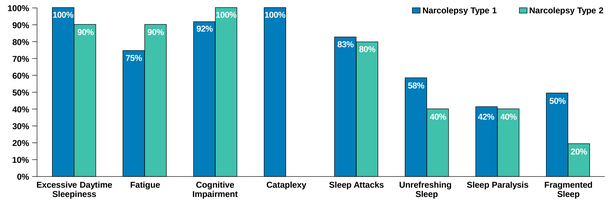
Figure 2. Time from Symptom Onset to Narcolepsy Diagnosis



- Symptom onset occurred most often in adolescence (n = 10, 45%) or early childhood (n = 6, 27%) among patients overall
- Initial symptoms typically included EDS (NT1 = 83%; NT2 = 80%), fatigue (NT1 = 42%; NT2 = 30%), oversleeping (NT1 = 33%; NT2 = 20%), sleep attacks (NT1 = 35%; NT2 = 50%), insomnia (NT1 = 35%; NT2 = 20%), and cataplexy (NT1 = 42%)
- 55% of patients overall (n = 12) reported that it took over 10 years for a diagnosis of NT1 or NT2 from symptom onset
- Half of participants (n = 11, 50%) described being misdiagnosed prior to their diagnosis of NT1 or NT2; misdiagnoses included depression (27%), sleep apnea (18%), and attention-deficit/hyperactivity disorder (9%)
- While seeking a diagnosis, participants reported visiting different types of healthcare professionals, including sleep specialists (NT1=7; NT2=4), neurologists (NT1=3; NT2=3), pulmonologists (NT1=2; NT2=2), psychiatrists (NT1=2; NT2=0), and primary care physicians (NT1=2; NT2=2)

FREQUENCY OF SYMPTOMS

Figure 3. Frequency of Symptoms Identified by Participants

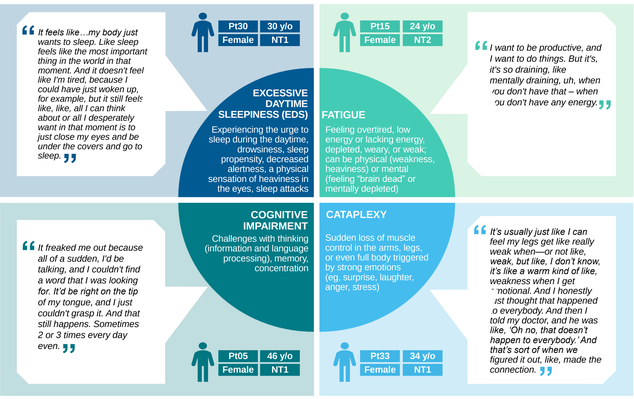


- Almost all participants (n = 21; 95%) described experiencing EDS
  - Over three-fourths of participants (n = 18; 82%) reported sleep attacks, which tended to be intense, sudden, and uncontrollable
- 82% of participants (n = 18) reported fatigue, frequently describing it as a feeling of low or depleted energy that left them constantly "exhausted," "dragging," and "drained" during the day
- Nearly all participants (n = 21; 95%) described cognitive impairments, such as feeling "slow" or "foggy"
  - More than half of participants described lapses in short-term memory (NT1 = 9; NT2 = 5) and difficulties with focus and attention (NT1 = 5; NT2 = 7); nearly half described trouble thinking through and processing information (NT1 = 6; NT2 = 3)
- All participants with NT1 reported experiencing cataplexy, commonly triggered by strong emotions/reactions such as laughter (n=6; 50%), anger (n=5; 42%), stress (n=5; 42%), and surprise (n=3; 25%)
  - Almost all participants with NT1 (n = 11; 92%) described experiencing cataplexy within a specific body part, such as the legs, hands, head, neck, or face
  - One-third of NT1 participants (n = 4; 33%) described full-body cataplexy, often resulting in them collapsing to the ground
  - Frequency of cataplexy varied, ranging from a couple times a year to weekly or daily
- For those that could identify their most bothersome symptom, fatigue was the most frequently cited (NT1 = 5; NT2 = 4), followed by EDS (NT1 = 5; NT2 = 2), and cognitive impairment (NT1 = 2; NT2 = 1)
  - Those who selected fatigue described it as constant, severe, and debilitating

FROM THE PATIENT PERSPECTIVE

Figure 4. Patient-Reported Symptoms: In Their Own Words

The 4 most common symptoms were EDS, cognitive impairment, fatigue, and cataplexy (NT1 = 100%).



Study Limitations

- The rare nature of narcolepsy resulted in difficulty recruiting study participants
  - Despite this, a saturation analysis suggested saturation of concepts was reached for NT1 and NT2 participants and additional interviews would likely not have yielded new information
- Participants included in this study were mostly female, White, and under 40 years of age
  - It is unclear if results would have been different with a more diverse sample with respect to gender, race/ethnicity, and age
- Confirmation of diagnosis was encouraged but not required; confirmation was provided by 4 participants with NT1 and 8 participants with NT2

CONCLUSIONS

- Results of this study provide a comprehensive description of the patient experience of narcolepsy and highlight the often long and complicated journey these individuals experience while seeking a diagnosis for their varied symptoms
  - Most patients reported that they had been misdiagnosed since the onset of their narcolepsy symptoms, and it took the majority of patients over 10 years to receive a diagnosis of NT1 or NT2
- The rich descriptions provided by participants for each symptom have contributed to a deeper understanding of how each may be experienced in patients' own words
  - From the patient perspective, NT1 and NT2 are characterized by a constant desire to sleep during the day; challenges with focus, concentration, and memory; and overwhelming and debilitating fatigue
  - Additionally, NT1 includes cataplexy that ranges in frequency, location, and emotional triggers

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DISCLOSURES

MJD and **WPW** are employees and stockholders of Alkermes, Inc. **MO'C**, **ML-C**, and **LTW** are employees of QualityMetric Incorporated, LLC.

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