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Making Sense of Narcolepsy

A Qualitative Exploration of How Persons with Narcolepsy Perceive Symptoms and Their Illness Experience

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Research Summary

Why was the research done?

This study was completed to fill the gap in our knowledge of the lived experience of persons living with narcolepsy in Australia.

What were the key findings?

Our findings suggest a misalignment between persons with narcolepsy and the medical establishment around how narcolepsy symptoms are conceptualised. We found that persons with narcolepsy often perceive the severity of narcolepsy by the level of functional impairment rather than symptom frequency. For the first time, we also identified anticipated and internalised or 'self-‘ stigma as the main types of stigma that persons with narcolepsy often experience.

What does this mean for policy and practice?

Our findings shed light on the perspectives, values, and preferences that persons with narcolepsy have around the management of care and service usage. They also highlight the substantial psychological comorbidity people with narcolepsy live with, presenting an opportunity for future research exploring the impact and possible development of stigma-related interventions.

Citation

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Making Sense of Narcolepsy: A Qualitative Exploration of How Persons with Narcolepsy Perceive Symptoms and Their Illness Experience

Abstract

Introduction: Understanding how persons with narcolepsy conceptualize symptoms, daily impact and illness experience is key to facilitating dialogue between patients and healthcare professionals. These concepts are usually explored from the perspective of healthcare professionals/researchers and rarely from the perspective of those with narcolepsy.

Methods: 127 self-reported persons with narcolepsy were recruited from an Australian patient support group. A short demographic survey was completed. All agreed to participate in a subsequent 1:1 semi-structured interview. Saturation was reached after 24 interviews (mean age = 33 years (SD 11) with 44% reporting cataplexy). A multidisciplinary team of researchers/clinicians analyzed interview transcripts using thematic analysis.

Results: Participants perceived physical fatigue, sleepiness, and two separate experiences of 'falling asleep/sleep attacks' as distinct symptoms rather than a multidimensional construct (i.e. excessive daytime sleepiness). We also identified two experiences of cataplexy, one triggered by acute emotion and another by a stressor. Participants determined their narcolepsy to be 'well-managed' by the level of functional impairment rather than the frequency of any symptom. Almost all participants described experiencing anticipated stigma and internalized or 'self-' stigma, likely stemming from societal devaluation of sleep and the conflation of sleepiness with laziness.

Conclusion: Descriptions of common symptoms often differed between participants and the existing literature. These differences likely impact patient-physician communication, with both parties utilizing the same terminology to communicate different concepts. The characterization of stigma in narcolepsy presents opportunities for future research exploring the impact and possible development of interventions to reduce the substantial psychological comorbidity in persons with narcolepsy.

Keywords: Narcolepsy, stigma, person-centred care, pathophysiology, validity, functional capacity, impairment, qualitative

1. Introduction

Identifying, diagnosing and assessing treatment efficacy in narcolepsy relies heavily upon the subjective experience of symptoms [1,2]. Often, the symptom experience is described from the clinical perspective of healthcare professionals rather than from the perspective of those living with narcolepsy. Narcolepsy is a rare sleep disorder characterized by several core symptoms, including excessive daytime sleepiness (EDS), cataplexy, hypnagogic hallucinations, sleep paralysis and disrupted nocturnal sleep [3]. Cataplexy is a key symptom of narcolepsy, described as a temporary loss of muscle tone, usually in response to positive emotion [3]. Identification and assessment of 'well-managed' cataplexy is almost exclusively measured subjectively using diaries to capture frequency of attacks, with objective testing rarely used [1]. The presence of cataplexy differentiates between the two types of narcolepsy: narcolepsy with cataplexy (narcolepsy type 1; NT1) and narcolepsy without cataplexy (narcolepsy type 2; NT2)[2].

EDS is another key symptom of narcolepsy. However, it is a non-specific term describing various phenomena related to sleepiness (e.g. sleep attacks, involuntary napping, difficulty sustaining attention). While these terms are used in clinical practice and across the literature, it is unclear whether persons with narcolepsy (PwN) use these terms in a similar way to healthcare professionals or even attribute the same meaning to these terms. Understanding how PwN conceptualizes symptoms is essential for facilitating dialogue between patients and healthcare professionals, ensuring that the needs, concerns and impact associated with narcolepsy can be communicated and addressed [4].

There is substantial evidence showing narcolepsy has an adverse impact on health-related quality of life, long-term disability, and absenteeism and is associated with poor socioeconomic and psychosocial outcomes [5-9]. Much of this research comes from European or US narcolepsy populations, with little known of the impact of narcolepsy on other populations. In Australia, we know little of the impact narcolepsy has on daily life, nor whether the healthcare system is meeting the needs and concerns of PwN. Australia offers universal healthcare and welfare and disability support programs, yet many of the narcolepsy treatments considered first-line internationally are not registered for use or considered second-line, likely affecting illness trajectory. At a recent government-mediated stakeholder meeting, Australians with narcolepsy made written submissions detailing their concerns with healthcare and unmet needs. The final report and policy recommendations from this meeting addressed healthcare infrastructure and resources [10]. Conversely, a document analysis of the written submissions by PwN and their family/carers found they were primarily concerned with unmet psychological needs, access to government services and treatment, and knowledge of healthcare professionals [11]

This study explores these issues in further detail, including the needs, concerns and challenges faced by PwN when navigating the Australian healthcare system.

Specifically, we sought to gain an in-depth understanding of the following:

1. how persons with narcolepsy describe symptoms that impact their daily life, and,
2. the needs, concerns, barriers, and facilitators to care faced by those living with narcolepsy in Australia.

2. Methods

2.1 – Ethics and design

The design of this study was a short cross-sectional survey and a 1:1 semi-structured interview that was analyzed thematically [12]. Ethics approval was granted by The University of Sydney Human Research Ethics Committee (reference 2021/110). The consolidated criteria for reporting qualitative research (COREQ) checklist was also used [13] (Supplementary A). A qualitative semi-structured interview schedule was developed by authors AS and JC (Supplementary B), informed by the findings of a previous thematic analysis of narcolepsy patient and family/carer submissions made to a public stakeholder meeting on the current state of sleep health in Australia [11].

2.2 – Recruitment and consent

Flyers, emails, and social media posts were used to recruit potential participants. A patient advocacy group, 'Narcolepsy Support Australia', assisted with recruitment by distributing these materials via their social media account. To be included, potential participants needed to live in Australia and have self-reported receiving a diagnosis of narcolepsy by a registered sleep specialist. Participants had the opportunity to review the participant information and consent form and ask questions before giving informed consent. Interviews were conducted using a consecutive and iterative approach, with purposive sampling [14] used to recruit a diverse sample of NT1/NT2, male/female experiences.

2.3 – Survey questions

Participants completed a short survey that collected demographic and other information about symptoms and delay in diagnosis. Specifically, participants indicated whether they experienced cataplexy brought on by emotion to assign them to NT1 and NT2 subtypes (with both subtypes included). Participants were also asked whether they were interested in participating in a subsequent one-hour semi-structured interview.

2.4 – Interview Procedure

One interviewer (AS) conducted semi-structured interviews during COVID restrictions from 30/06/2021 – 14/10/2021 using Zoom, an online meeting platform. Both audio and video recordings were collected, with field notes written throughout all interviews. Data saturation was determined to have occurred when no new needs, concerns, or barriers to care were mentioned or different perspectives were given on existing topics across three consecutive interviews. Saturation was based on field notes and decided by three authors (AS, JC, NG) during fortnightly meetings.

2.5 – Data analysis

Survey data were analyzed using statistical software package SPSS (version 25.0). Interview recordings were transcribed using the NVivo Transcription Service, then anonymized and checked for accuracy by an author (AS) and research assistant (CK). Data was interpreted thematically using the established six-step process of qualitative analysis [12], as it provides a flexible method of analyzing and interpreting substantial amounts of qualitative data. Five authors from diverse backgrounds (e.g. qualitative research, medicine, pharmacy and lived experience) participated in the analysis (AS, JC, NT, DN, AM). The six steps include: (1) Become familiar with the data: Five randomly selected transcripts were allocated to each team member to familiarise themselves with the ideas and concepts discussed in the interviews. Insights, concepts and ideas identified by the analyzing team were recorded; (2) Generate initial codes: the research team initially explored a sub-sample of data by making comments in the participants' own words in a Microsoft Word document of the de-identified transcripts to develop a preliminary coding framework; (3) Search for themes: Open coding was conducted using NVivo 12 Software by one of the analysis team members; 4 & 5) Review and define themes: the themes in the coding framework continued to be collaboratively refined and named through an iterative process of reading, coding, reflection and discussion in fortnightly team meeting until all significant parts of the data had been considered. A codebook was collaboratively developed, which included sub-themes and overarching themes. All interviews were subsequently coded by author XX, with 20% double coded by author NT to check for reliability. The collaborative approach to analysis supported reflexivity as it encouraged comparisons and sharing of diverse perspectives the research group offered with their various backgrounds and lived experiences [15]; and 6) Write-up: the results were written up and reviewed by all authors.

3. Results

3.1 – Description of participants

We recruited 127 participants who self-reported receiving a diagnosis of narcolepsy from a registered healthcare professional. All participants indicated they would like to take part in subsequent interviews. Saturation was reached after twenty-four semi-structured interviews (ranging from 34 min 43s to 68 min 13s in length, averaging 51 min 01s). Half of the participants reported experiencing cataplexy, while 70% reported experiencing a "sleep attack". Approximately 60% reported experiencing symptoms in adolescence, yet only 23% received a diagnosis before turning 18. Further, 22% reported a delay in diagnosis of > 10 years from symptom onset.

Table 1: Demographics of interviewed participants

Demographic	Interviewed participants (n = 24)
Age – mean (range, SD)	33.4 (22 – 58, 10.8)
Sex – Female (n, %)	15 (63%)
Resides in capital city – n (%)	16 (67%)
Symptoms	
Cataplexy – n (%)	10 (42%)
Sleepiness *	
Sleep attack – n (%)	16 (70%)
Fatigue – n (%)	22 (96%)
Never rested – n (%)	16 (70%)
Symptom onset *	
> 18 years of age	14 (61%)
≤ 18 years of age	9 (39%)
Age of diagnosis *	
> 18 years of age	5 (23%)
≤ 18 years of age	18 (77%)
Delay in diagnosis from onset of symptoms *	
< 3 years	7 (30%)
Between 3 - 10 years	11 (48%)
> 10 years	5 (22%)

* n = 23

3.2 – Thematic analysis

Our analysis identified four themes: 1) The symptom experience of narcolepsy – perspectives of excessive daytime sleepiness and cataplexy; 2) Making sense of the illness experience – perspectives of identity, daily impact, and the label of narcolepsy; 3) Making sense of long-term care: narcolepsy management following diagnosis; and 4) Making sense of the perception of narcolepsy – how do people with narcolepsy and others perceive the disorder.

3.2.1 – Theme 1: Making sense of the symptom experience of narcolepsy – perspectives of excessive daytime sleepiness and cataplexy

3.2.1.1 – Subtheme 1: Excessive Daytime Sleepiness – perceptions of a multidimensional construct

All participants reported experiencing symptoms that are usually grouped under the term Excessive Daytime Sleepiness (EDS) every day, with three broad components described: 1. Fatigue / physical lack of energy, 2. The feeling of being sleepy / sleepiness, and 3. the act of falling asleep (often called a "sleep attack"). Each component was perceived as a distinctly separate construct from other aspects of EDS (figure 1). Each component was also associated with varying degrees of functional impairment (ranging from minimal to substantial impact on daily life, e.g. an inability to work). Participants also used the term 'tiredness' interchangeably used to describe either fatigue or sleepiness.

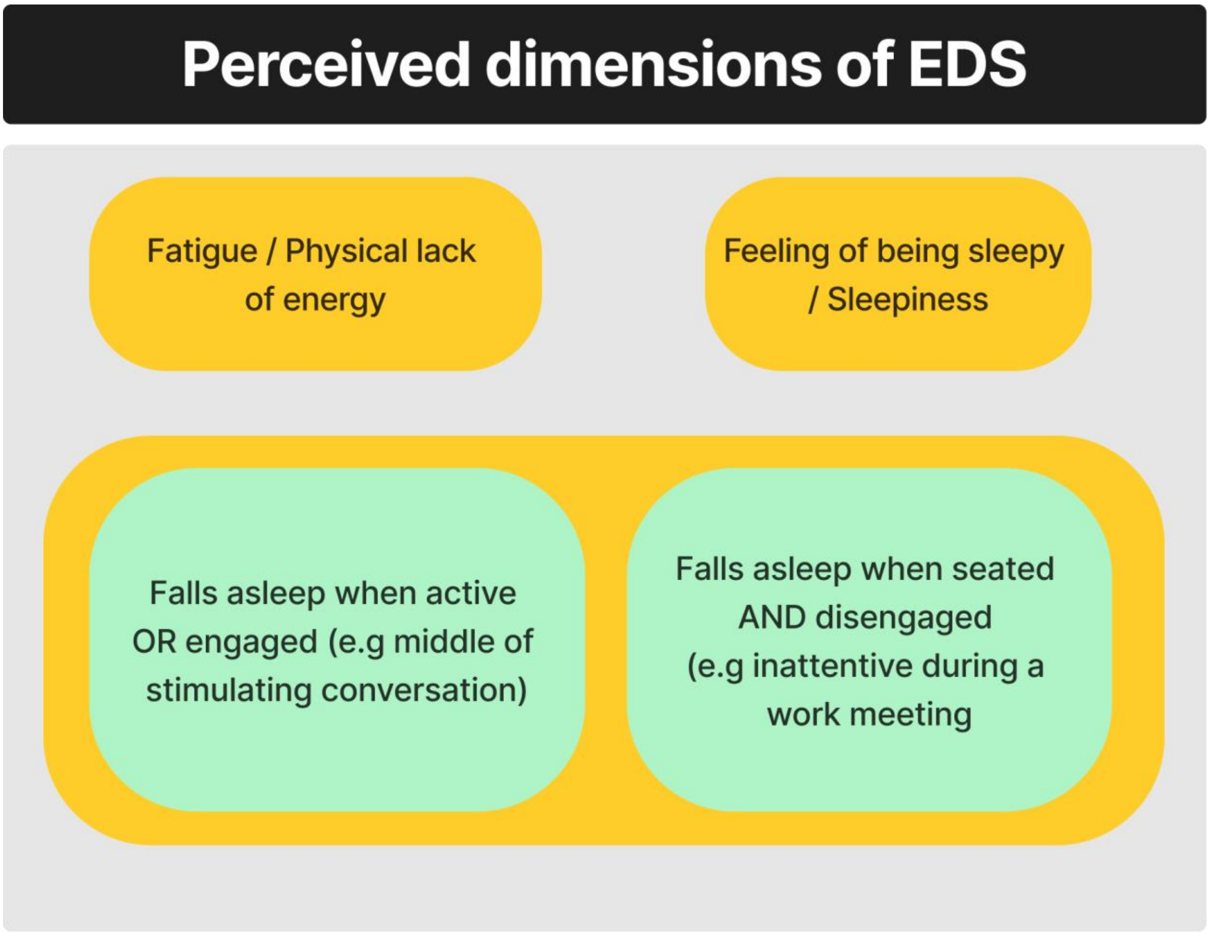

Figure 1: How persons with narcolepsy perceive the symptom Excessive Daytime Sleepiness (EDS). The yellow boxes represent individual symptoms that were explicitly differentiated between by participants. These symptoms are often considered related to one another or unidimensional by the medical establishment. The green boxes represent two distinct symptom experiences that are often referred to as a 'sleep attack'.

Fatigue was described as exhaustion and a lack of physical energy that was explicitly separate from sleepiness and falling asleep. Participants described fatigue as synonymous with physically feeling "drained", with some describing it as the most functionally impairing component of EDS.

"I often also get very fatigued; drained and physically can't do much" [P24, NT2]

Sleepiness was described as a *"crushing need to sleep"* [P18, NT2] and often referred to as a physical pressure. This feeling was sudden and acute for some, while others described it as a dull, day-long physical pressure and a "*a need to be prone and close your eyes and lie down*" (P18, NT2). This symptom was perceived to contribute to poor concentration and cause 'brain fog'.

Two distinct accounts of the act of falling asleep were described. The first would only occur when the participant was seated and was not actively engaged, often described as situations where they were bored, unfocused/inattentive, or performing a menial task. These participants explicitly stated that they did not fall asleep while standing or actively engaged in a task, as they could 'overcome' or' push through' their sleepiness in these situations (Box 1).

The other account of the act of falling asleep was described by fewer people (n=4) as a sudden, acute onset of sleep that could occur despite being active, physically moving, standing, or mentally engaged in a task (e.g. socializing, walking, or when the participant was talking "*mid-conversation with my partner*" (P6, NT1)). This was preceded by a sudden, acute feeling of "sleepiness" or sleep pressure. One participant said that if they avoided their scheduled nap and tried to "*push through"* (P22, NT1), a sleep attack would eventually occur regardless of the activity being performed. Another described this occurring multiple times a day, with family and friends able to recognize external signs of when this episode would occur, where they would *"see my eyes glaze over and he can spot the signs a mile off"* (P6, NT1). The act of falling asleep when 'active or engaged' was only described by participants self-reporting cataplexy associated with acute positive emotion. Participants who described their cataplexy as "*not full-blown cataplexy*" (P2, NT1) or "*not dropping completely with, you know, high, like, emotions*" (P9, NT1) reported that they did not fall asleep while actively engaged.

Perhaps most important was the language used to describe these symptoms. Participants clearly distinguished between two distinct acts of falling asleep when describing their symptoms. However, terms like "sleep attack" and "falling asleep mid-task" were used interchangeably to describe their experiences, even by persons who experienced both types. While some participants only experienced one of the two described acts of falling asleep, they were also aware that others with narcolepsy had a different experience (box 1).

> *"They were type 1 and they just fell asleep throughout the day randomly, and I was like, oh, I don't just fall asleep randomly throughout the day. I'm like, I just get tired real quick when I'm not doing anything, it's not like I just drop off involuntarily. I'm like, it's a…it's something that just is, um, I just am prone to getting tired very quickly when not doing anything. It's like, it's definitely different.*" [P3, NT2]

Box 1: A participant with narcolepsy type 2 describing the perceived differences between symptoms experienced by someone with narcolepsy type 1

3.2.1.2 – Subtheme 2: Lived experience of cataplexy – differences in triggers

There was variability in the descriptions and experiences of participants who self-reported cataplexy (figure 2). Six of the ten participants self-reporting NT1 described their cataplexy as general muscle weakness, "*slurring*" [P9, NT1], tongue protrusions and knee-buckling that was infrequent, intermittent, or not impactful on daily life. Three of these also recalled uncertainty around whether their experience met the clinical definition of cataplexy, despite identifying with this label:

"*I don't feel like I've had [classic cataplexy] episodes*" [P9, NT1].

Another person described a loss of consciousness when describing their experience. Four participants reporting cataplexy (40%) described it as being triggered by acute emotion, with laughter as a specific trigger. All four described experiencing both full and partial attacks (i.e. cataplexy resulting in full body paralysis vs momentary weakness in limbs/face) that had caused them physical injury or embarrassment in the past. Notably, despite pharmacological management, these participants described their cataplexy as a permanent and persistent symptom, one where they needed to be constantly mindful of their emotions and potential triggers, with the participant constantly vigilant of an attack. This contrasted with the experience of the six participants who did not experience cataplexy associated with acute emotion and who described their attacks as single, intermittent or one-off events driven by chronic triggers (i.e. stress), that the cataplexy had resolved itself or not occurred for extended periods (e.g. > five years).

Several participants also used the term "cataplexy" for symptoms not consistent with the clinical criteria for cataplexy. In some cases, participants used the term to describe what the literature refers to as a "sleep attack", with cataplexy perceived as a version of falling asleep:

"I could fall asleep at the shops. Um, for me, though, I can fight it. I feel some people, some people can't, and that's where the cataplexy comes in" [P14, NT2]

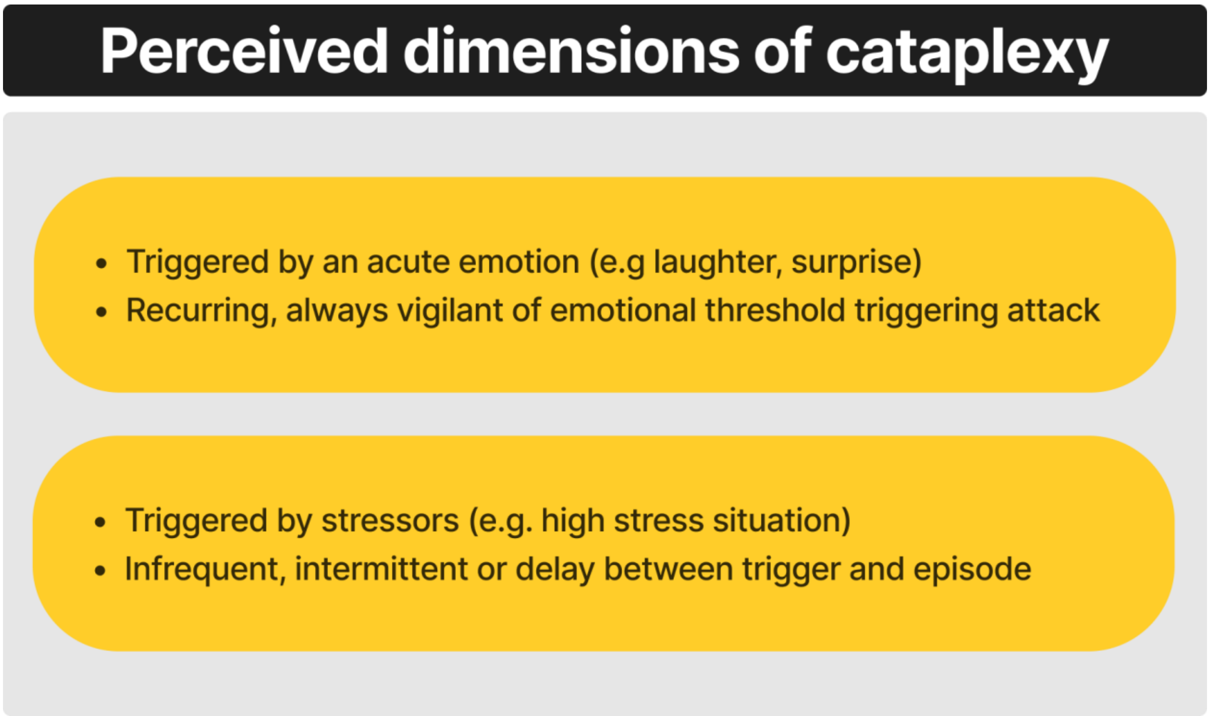

Figure 2: How persons with narcolepsy perceive the symptom cataplexy (complete and partial episodes). The yellow boxes represent the two distinct experiences of cataplexy, as described by participants.

3.2.2 – Theme 2: Making sense of the illness experience – perspectives of identity, daily impact, and the label of narcolepsy

3.2.2.1 – Subtheme 1: "You may as well have something that's completely different than what I do" – Differences in illness identity

Participants distinguished their illness identity from others diagnosed with narcolepsy based on their experience of the two symptoms above: cataplexy and 'the act of falling asleep'. Almost all participants without cataplexy described NT1 as more severe and functionally impactful and perceived this as a separate illness experience from their own. Some (n=8) described themselves as "*thankful*" [P4, NT2] and "*lucky*" [P14, NT2] they did not have cataplexy, viewing their illness experience of NT2 as more favourable. This view was shared by participants with cataplexy not triggered by emotion, who described their experience of cataplexy as not impairing or limiting, where they have "*the blessing of not having full-blown cataplexy*" (P2, NT1). The small number of NT1 participants self-reporting cataplexy triggered by positive emotion similarly perceived their illness experience as different from NT2 on the basis of increased functional impairment and their understanding of narcolepsy physiology:

"I think that plays out completely different than it does for a narcoleptic who just has the sleepy side of things. It's a completely different mechanism. And I think that not having [orexin], I don't know that it's just about fighting the urge to sleep. There is so much more to it than that. It plays out in everything." [P6, NT1]

Participants also distinguished their illness experience depending upon whether they experienced 'the act of falling asleep' while active and engaged. The majority who did not experience this aspect of EDS described frustration that the public perceived narcolepsy as falling asleep at any time, which did not align with their own experience. Many felt this contributed to the misunderstanding and confusion around narcolepsy and its impacts:

"It's the trope that I'll fall asleep while standing up" [P2, NT1]

Only a few participants did not limit descriptions of their illness experience to cataplexy or sleep attacks. Instead, these participants defined their illness experience of narcolepsy as extending beyond the typical symptoms associated with narcolepsy, attributing a wider range of symptoms, functional impacts, and other comorbidities to their experience of narcolepsy label.

"There is a lot more to the surface [of narcolepsy] than just the four main symptoms, I guess, it's definitely a lot more than just that" [P1, NT2]

3.2.2.2 – Subtheme 2: Diagnosis and the Spectrum of Acceptance

Two distinct groups of participants were identified based on their acceptance of their diagnostic label. The first described their diagnosis as validating and embraced the label of narcolepsy as it provided answers to long-standing questions about health, unexplained symptoms or why they perhaps were not as functional as others:

"*More relief that we finally found out what it was and no more rushing around doctors*." [P19, NT1]

Conversely, the second group attributed negative connotations to the diagnosis, with some outright rejecting it, not wanting to be perceived as disabled or less functional. Others considered their diagnosis a mistake, with two describing their diagnosis as forced upon them by healthcare professionals. This was experienced by a group that was primarily defined by symptoms that fluctuated in severity or frequency or were now resolved:

"I literally didn't believe it. I did not believe it. I was like, nah, it must be some kind of mistake." [P11, NT2]

All participants attributed past experiences, such as poor performance in school, university, work or other life events, to their narcolepsy rather than themselves.

3.2.2.3 – Subtheme 3: Assessing well-managed narcolepsy by the daily impact of symptoms

Participants determined if their narcolepsy was well-managed by their level of functional impairment rather than symptom severity (i.e. a desire to be functional/productive at work rather than difficulties staying awake/feeling sleepy at work).

Most reported being able to work full-time. Participants that only fell asleep when seated and disengaged described a constant "battle" against sleepiness that persisted throughout the day that affected concentration and focus. Most were able to work full time and reported being able to "*overcome*" or "*push through"* symptoms by, for instance, remaining standing in meetings, choosing a role that kept them active, or having a scheduled nap, with few (n=2) describing symptom management as a *"frame of mind"* [P23, NT1]. Participants also described their medication regimes, with several describing it as optional:

"I don't really take my medication too much, but if I really do need to wake up, or like to focus, then I'll take my medication" [P1, NT2]

Those that fell asleep when active and mid-task described a constant fear of having an episode in public or without an appropriate or safe place to do so, with these episodes occurring regardless of activity. All three participants described difficulty finding or maintaining employment. Both groups described inadequate accommodations, with some describing themselves as seeking out bathrooms at work to *"nap in the toilet"* [P9, NT1].

Participants also described how they manage their fatigue. Most prioritized work or career at the cost of leaving adequate energy for basic self-care activities or carer responsibilities. Participants described that they "*can't do all those things and that something has to give*" [P24, NT2], often resulting in conflict within relationships as family members or partners had to take on the additional burden. Energy levels were managed by keeping strict routines and structuring daily activities to account for their reduced capacity. Self-preservation behaviour was described, with participants often becoming upset, distressed, or defensive when their routine was interrupted by unexpected or external circumstances, as this would exacerbate the severity and frequency of symptoms.

The few with cataplexy triggered by positive emotion (n=4) suggested this symptom often exacerbated other symptoms of narcolepsy, including increased fatigue and episodes of falling asleep:

"It's been really involved and full on like emotionally speaking, my functionality, that'll wipe me for the rest of the day" [P6, NT1].

The impact of cataplexy went further than the episode itself, with all describing the psychological impact of not being able to experience or regulate emotion. This affected the participants' ability to engage with others, maintain personal relationships and socialize, with a constant need to maintain vigilance over experiencing triggering emotion described:

"it's so scary having to like be sure that you don't accidentally trigger the cataplexy" [P17, NT1].

One participant spoke of his school experience where friends would try to trigger a cataplexy attack, describing the psychological impact of choosing between his friendship group or his safety.

3.2.3 – Theme 3: Making sense of long-term care – narcolepsy management following diagnosis

3.2.3.1 – Subtheme 1: Sourcing information and support about narcolepsy

Digital media was a primary source of information about narcolepsy and upcoming treatments. Websites, online support groups, social media, online forums (i.e. Narcolepsy/Idiopathic hypersomnia Reddit community) and digital peer-reviewed journal articles were all described. Few (n=4) mentioned their specialist as their primary source of information, with most describing themselves as more knowledgeable about narcolepsy.

Three specific areas were identified where information was considered lacking. Firstly, participants knew of few educational resources they could give to workplaces or schools/universities to explain the impact of narcolepsy or potential accommodations. The second related to the eligibility and the application process for government support services (i.e., National Disability Insurance Scheme (NDIS)), with several participants unsure whether narcolepsy was considered a disability or how to apply:

"I kind of feel like we're a bit of a, you know, other group, like you're not recognized" [P5, NT2]

Lastly, women with narcolepsy described limited information about the safety of medications for narcolepsy, with several reporting they knew medications like modafinil were not suitable for pregnant or breastfeeding women. Participants spoke of a top-down approach taken by their treating specialist where they were told they were not allowed to continue their prescribed medication. These same participants said the ideal situation would have been sitting down and discussing the risks and benefits of continuing treatment with their treating doctors. A unique set of psychological challenges resulted from this, where women feared choosing between maintaining their functional status and independence or having a child.

"I didn't find any resources anywhere, and I think I'm a pretty good [at googling] about what narcolepsy and pregnancy means. So, I had no idea if having children or being pregnant was going to screw with me" [P7, NT2]

3.2.3.2 – Subtheme 2: The relationship with healthcare professionals

Participant satisfaction with their treating sleep specialists varied across interviews. A steady, long-term specialist was perceived as key to instilling trust between patient and specialist, with some participants seeing the same specialist for >5 years. For most others, a misalignment in the treatment priorities and a perceived lack of understanding by clinicians of the whole person impact of narcolepsy was described. This misalignment affected the patient-specialist relationship, where many perceived their doctor as a passive provider of medication rather than an active decision-maker involved in managing their narcolepsy:

"my ongoing relationship with him is the script, to the extent that I want the script, and he would check-in, like as a 'high-level' like, are you okay?" [P7, NT2].

While some were satisfied with this approach, others perceived this as their specialist lacking knowledge and training specific to narcolepsy. Several participants attributed this to sleep medicine and respiratory medicine being combined under one speciality in Australia, with sleep specialists perceived as lacking training specific to non-respiratory sleep disorders. This had a reported impact on the management of narcolepsy through public hospital sleep clinics, where participants were not given a choice of physician they were referred to, with some describing referrals to physicians who specialized primarily in respiratory medicine rather than in sleep disorders:

"the specialist I see is a thoracic surgeon, an ear, nose and throat doctor because it's the [public hospital] sleep and respiratory unit" [P6, NT1]

Some participants brought their research and information to their specialists, with few specialists described as receptive to discussing the information. For others, their research was dismissed, which was particularly damaging to the patient-physician relationship.

Others described themselves as treatment-seeking rather than seeking out the expertise or knowledge of a specialist:

"shop around a fair bit to get medical professionals that are understanding or knowledgeable enough to, to sort of continue with my treatment" [P24, NT2]

3.2.4 – Theme 4: Making sense of the perception of narcolepsy – how do people with narcolepsy and others perceive the disorder

3.2.4.1 – Subtheme 1: What do others think of my narcolepsy? – others' perceptions of narcolepsy

In every interview, participants universally feared being caught asleep, shamed or *"being judged or misunderstood or um, I guess, being seen that I'm not capable"* [P5, NT2]. These negative sentiments were not directed towards narcolepsy itself (i.e. part of a stereotype of narcolepsy). Rather, these sentiments were directed towards attitudes to sleep, sleepiness, and napping, all conflated with laziness and being unproductive. Those closest to the participant, including family members and work colleagues, often held these negative sentiments. These individuals often trivialized or normalized the experience of the PwN by comparing it to their own experiences of fatigue and sleepiness or not considering these symptoms to be a medical condition:

"I'm not some tired piece of crap who can't stay awake. I'm genuinely fighting something" [P3, NT2]

The trivialization resulted in a breakdown of trust and confidence for some, with participants describing an unwillingness to communicate their healthcare needs or the impact of narcolepsy with others:

" I was like 'making it up' type of thing or like 'it wasn't real' type of thing. It definitely added some strain to the relationship, and it's definitely made me more secretive about my health problems" [P1, NT2]

Participants also described encountering negative perceptions towards narcolepsy treatments. Several medications used to treat narcolepsy were known to be misused as study aids amongst university students and as performance enhancers in the workplace, and participants thought they might be viewed as using them for similar purposes. Some healthcare professionals also perceived as stigmatizing narcolepsy treatments, with one participant describing they *"get that whole drug-o thing feeling [from my pharmacy]"* [P16, NT2] when collecting their monthly stimulant medication.

Several participants described specific instances where they felt discriminated against in the workplace, not based on the diagnostic label of narcolepsy but rather the symptoms or functional impairment (e.g. being late for work, falling asleep or mistakes made due to issues with concentration/brain fog). Participants also feared being discovered to have narcolepsy as they did not want to be perceived as less capable. Often, this fear intersected with the different experiences of sleep attacks, with participants concerned that employers might search the internet for information on narcolepsy and assume they would fall asleep mid-task and thus be perceived as a liability:

"Even though I'm all right, I'm fully medicated and it doesn't, you know, I can get through most of the day, they'll still go, yeah but when I googled you, you could fall asleep" [P22, NT1]

While most did not describe experiencing actual discrimination, almost all feared or assumed they would be discriminated against (i.e. anticipatory stigma). The fear of discrimination led some to engage in potentially unhealthy behaviours to try and stay awake, such as substantial consumption of caffeine and energy drinks in addition to stimulant medication, or one case, through pain:

"I keep a rubber band around my wrist, like just to flick, keep myself awake, drink cold water, wash my face, I'll even bite a lemon, that extreme" [P23, NT1]

3.2.4.2 – Subtheme 2: How is narcolepsy perceived by those living with it

Almost all participants appeared to have internalized their stigma, either agreeing with negative stereotypes around sleepiness and unproductivity and subsequently trying to distance themselves from the label of narcolepsy or hide their symptoms. This extended to participants who were amongst the most functional, including those employed full-time or who did not describe the substantial daily impact. Some felt shame stemming from being diagnosed with narcolepsy while others were even sympathetic to the idea that employers would discriminate against someone with narcolepsy, often describing the rationale behind such discrimination as understandable:

"I was literally just [pauses] not reliable. If I was my own employee, I would have sacked me." [P11, NT2]

Perceptions of narcolepsy also varied with levels of daily function. PwN who were more functional described the limited functionality of others with narcolepsy as by choice or a consequence of their own decisions:

"people seem to make excuses for themselves rather than help themselves out" [P2, NT1].

Participants also had negative perceptions of others with narcolepsy based on their symptom experience. Some of those with NT2 who described themselves as less functionally impaired perceived those with NT1 or persons that fell asleep while standing and mid-task as lazy or unproductive, with one NT2 participant describing someone with NT1 as *"dopey and like just constantly asleep like a human sloth. Whereas [my narcolepsy] was never like that."* [P8, NT2].

4. Discussion

To our knowledge, this is the first qualitative exploration of the symptom experience of narcolepsy from the perspective of persons with narcolepsy (PwN). Qualitatively, we found that persons with NT1 and NT2 described different experiences of symptoms and illness, identities, and levels of functional impairment. Participants distinguished between the two subtypes of narcolepsy, with some stigmatizing those less functional or with different symptom experiences than their own. Our results raise questions about whether NT1 and NT2 should be considered subtypes of a single disorder or two different disorders, as many PwN themselves appeared to consider these separate.

We found that PwN reported four distinct and distinguishable symptoms often grouped under the umbrella term 'EDS'. We also observed two distinct experiences of cataplexy. Each symptom carried varying degrees of functional impairment and impact on daily life and should be considered and measured as separate constructs to reflect the lived experience of narcolepsy (e.g. in PROMs). While umbrella terms like 'EDS' are standard in the sleep field, its use appears to lack the specificity to describe and convey the symptom experience. Further, ongoing use of shorthand could contribute to trivializing narcolepsy symptoms, as 20% of the general population is also purported to experience EDS [16]. There are also implications for narcolepsy clinical trials, as the choice of primary outcome measure for EDS is likely underpinned by a false assumption of content validity (e.g. outcome measure used to capture all aspects of EDS, as experienced by PwN [1,17]). It suggests a need to move away from selecting outcome measures for EDS in efficacy trials towards the assessment of each individual symptom. Not only would this allow healthcare professionals to create tailored treatment plans, but also the ability to better meet the needs and priorities of PwN.

There was also a lack of common language and terms that PwN could use to convey their symptom experience. Participants used "sleep attack" and "falling asleep mid-task" interchangeably to describe their symptom experience, where each person attributed their meaning to these terms (e.g. describing a sleep attack but calling this experience 'cataplexy'). The meaning prescribed to these terms often differed between subtypes of narcolepsy and from definitions given throughout the literature. Overall, there appears to be a discrepancy between the language used by the medical establishment and PwN when describing symptoms experienced, with much lost in translation [3,18]. For effective communication between patients and healthcare professionals, there needs to be a shared understanding and language to convey the subjective experience of symptoms, ensuring all relevant domains are defined [19]. It suggests the need for an agreed-upon, clearly defined language developed by PwN, healthcare professionals and researchers before we can effectively communicate the illness experience. Defining these symptoms is a necessary first step towards creating a valid patient-reported outcome measure and ensuring the validity of existing data collection systems that use these terms and explore these experiences.

We also found discrepancies between how 'well-managed narcolepsy' is perceived and measured. Most participants assessed the severity of their narcolepsy by their level of functional impairment rather than the frequency or severity of symptoms, which are the core of how clinicians and researchers measure treatment efficacy in trials. This implies that narcolepsy is not just a disorder to be treated but a functional disability to be managed. Previous studies support such an approach, with self-reported sleepiness and global improvement strongly correlated with measures of function and health-related quality-of-life rather than objective measures of sleepiness [20]. However, clinical guidelines for the management of narcolepsy appear to reflect the opposite, with symptom frequency prioritized over the whole person's functional impact of the disorder [21,22]. To bridge this gap and assess whole-person function, simple analogies that help PwN describe their perceived energy levels and ability to perform daily tasks can be valuable tools for healthcare professionals (e.g., spoon theory or battery analogy – see SUPPLEMENT C [23]). In the interim, healthcare professionals can use these to help understand how patients convey the functional impact of their disease. While useful, our results suggest a need for patient-reported outcome measures that assess narcolepsy using endpoints considered more meaningful to patients.

Another important finding from this study was the characterization of the stigma that PwN reportedly experienced [9]. Much of this stigma appeared directed towards the symptoms of narcolepsy rather than the diagnostic label of 'narcolepsy' itself. These symptoms included sleepiness, falling asleep and napping, and were frequently conflated with failure, laziness and being unproductive. Anticipated stigma was amongst the primary type of stigma experienced by PwN. In the context of living with narcolepsy, anticipated refers to the extent a person with narcolepsy believes other people would devalue/distance themselves if others found out they were overly sleepy, fell asleep more often or required regular naps [24]. This stigma likely intersects with Western societal values around productivity and the devaluation of sleep [25,26]. Living in a society that denigrates sleepiness, falling asleep and napping likely result in the internalized or 'self- 'stigma observed in almost all participants [24].

Both anticipated and internalized stigma are often experienced by people who have a *concealable stigmatized identity*: identities or attributes that can be hidden from others but are socially devalued and negatively stereotyped (e.g. mental health, sexual orientation, persons with HIV-AIDS) {Quinn, 2014 #938}. The experiences of PwN have striking resemblances with others who have a concealable stigmatized, with narcolepsy status perhaps considered a concealable stigmatized identity. These findings are important as anticipated and internalized stigma has been shown to strongly predict psychological distress in populations with a concealable societal devalued identity [24], likely contributing to the high prevalence of depression, anxiety, suicidal ideation, and other psychological comorbidities observed in narcolepsy [28-30]. Considering those with narcolepsy have a biological propensity to fall asleep, the chronic stress of trying to hide their symptoms or being 'outed' likely contributes to the psychological comorbidity of narcolepsy [27-29]. With much of this stigma described as experienced in a workplace setting, it also highlights a broader socio-legal implication related to occupational health and safety. If employees with narcolepsy anticipate stigma and hide their symptoms, they may decide not to disclose this to their employer, despite having a condition that might increase the probability of an accident during work hours due to a sleep attack or cataplexy. One possible solution would be the creation of jurisdiction-specific educational programs or information packs for employers that include details about narcolepsy symptoms and appropriate accommodations (e.g. including work-from-home practices, allowing to stand during meetings)[31].

4.1 – Strengths and Limitations

The first author (AS) is a lived-experience researcher diagnosed with narcolepsy type 1, which may be a strength, as it allowed participants to engage with a fellow community member, sharing insight and experiences that they otherwise may not have shared with someone without narcolepsy. Conversely, AS acknowledges that this may have biased the direction of the interviews and inferences, given his experience with narcolepsy and the contention raised between distinguishing between subtypes. Using a reflexivity diary and having a large research team with diverse experiences and backgrounds (e.g., healthcare professionals, psychologists, pharmacists, and other academics from outside of sleep research) helped challenge preconceived ideas and encouraged AS to remain aware of potential biases. Another strength was the substantial response of potential participants to the screening survey. It enabled us to purposively sample based on narcolepsy subtype and sex, increasing the possibility that the experiences described were indicative of the narcolepsy community at large. Limitations of this study include that all interviews were conducted in English and therefore lacked representation of non-English-speaking PwN who may have different cultural perceptions of sleep and experiences of an English language-dominated health care system, which is essential considering the multicultural population of Australia. Our study was also limited to those self-reporting a diagnosis of narcolepsy. No objective measures or clinical data were collected or a way to confirm the diagnosis, perhaps contributing to the heterogeneity observed in our cohort.

5. Conclusion

Our findings suggest the symptom experience of narcolepsy is more heterogeneous than what is described in diagnostic manuals and the literature, where participants attributed their own meaning and experience to commonly used terminology (i.e sleep attack). The discrepancy in terminology could affect patient-physician communication, with both parties utilizing the same terminology to communicate different concepts. It could also have implications for research that utilize PROMs to capture symptom severity/experience. Further work is also needed to bridge the gap between the healthcare needs and persons with narcolepsy and healthcare professionals, including the provision of care that addresses functional capacity and impairment rather than symptom treatment. Having characterized the stigma experienced by PwN as both anticipated stigma and internalized or 'self-'stigma, our findings presents an opportunities for future research exploring the impact and possible development of tailored interventions to reduce the substantial psychological comorbidity in persons with narcolepsy.

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Competing interests

Aaron Schokman is a member of several narcolepsy patient support groups. No other author has any conflicts of interests to declare.

Data Availability Statement

The data underlying this article cannot be shared publicly due to privacy and ethical concerns of individuals that participated in the study.

References

1. Schokman A, Bin YS, Naehrig D, Cheung JM, Kairaitis K, Glozier N. Evaluation of psychometric properties of patient-reported outcome measures frequently used in narcolepsy randomized controlled trials: a systematic review. *Sleep.* 2022:zsac156.
2. Sateia MJ. International classification of sleep disorders. *Chest.* 2014;146(5):1387-1394.
3. Bassetti CLA, Adamantidis A, Burdakov D, et al. Narcolepsy — clinical spectrum, aetiopathophysiology, diagnosis and treatment. *Nature Reviews Neurology.* 2019;15(9):519-539.
4. Health UDo, Services H. Guidance for industry-Patient-reported outcome measures: Use in medical product development to support labeling claims. *http://wwwfdagov/Drugs/GuidanceComplianceRegulatoryInformation/Guidances/defaulthtml.* 2009.
5. Thorpy M, Morse AM. Reducing the clinical and socioeconomic burden of narcolepsy by earlier diagnosis and effective treatment. *Sleep Medicine Clinics.* 2017;12(1):61-71.
6. Black J, Reaven NL, Funk SE, et al. The Burden of Narcolepsy Disease (BOND) study: healthcare utilization and cost findings. *Sleep medicine.* 2014;15(5):522-529.
7. Flores NM, Villa KF, Black J, Chervin RD, Witt EA. The humanistic and economic burden of narcolepsy. *Journal of Clinical Sleep Medicine.* 2016;12(3):401-407.
8. Plazzi G, Clawges HM, Owens JA. Clinical characteristics and burden of illness in pediatric patients with narcolepsy. *Pediatric Neurology.* 2018;85:21-32.
9. Kapella MC, Berger BE, Vern BA, Vispute S, Prasad B, Carley DW. Health-related stigma as a determinant of functioning in young adults with narcolepsy. *PloS one.* 2015;10(4):e0122478.
10. Bedtime Reading: Inquiry into Sleep Health Awareness in Australia. In: House of Representatives Standing Committee on Health ACaS, ed: Parliament of the Commonwealth of Australia; 2019.
11. Schokman A, Glozier N, Aji M, Bin YS, Kairaitis K, Cheung JM. How patient centric is health policy development? The case of the Parliamentary Inquiry into Sleep Health Awareness in Australia. *Australian Health Review.* 2022;46(2):233-243.
12. Braun V, Clarke V. Using thematic analysis in psychology. *Qualitative research in psychology.* 2006;3(2):77-101.
13. Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *International journal for quality in health care.* 2007;19(6):349-357.
14. Kvale S, Brinkmann S. *Interviews: Learning the craft of qualitative research interviewing.* sage; 2009.
15. Barry CA, Britten N, Barber N, Bradley C, Stevenson F. Using reflexivity to optimize teamwork in qualitative research. *Qualitative health research.* 1999;9(1):26-44.
16. Swanson LM, Arnedt JT, Rosekind MR, Belenky G, Balkin TJ, Drake C. Sleep disorders and work performance: findings from the 2008 National Sleep Foundation Sleep in America poll. *Journal of sleep research.* 2011;20(3):487-494.
17. Mokkink LB, De Vet HC, Prinsen CA, et al. COSMIN risk of bias checklist for systematic reviews of patient-reported outcome measures. *Quality of Life Research.* 2018;27(5):1171-1179.
18. Khatami R, Luca G, Baumann CR, et al. The European Narcolepsy Network (EU-NN) database. *Journal of sleep research.* 2016;25(3):356-364.
19. Kluger BM, Krupp LB, Enoka RM. Fatigue and fatigability in neurologic illnesses: proposal for a unified taxonomy. *Neurology.* 2013;80(4):409-416.

20. Weaver TE, Mathias SD, Crosby RD, et al. Relationship between sleep efficacy endpoints and measures of functional status and health-related quality of life in participants with narcolepsy or obstructive sleep apnea treated for excessive daytime sleepiness. *Journal of sleep research.* 2021;30(3):e13210.
21. Bassetti CL, Kallweit U, Vignatelli L, et al. European guideline and expert statements on the management of narcolepsy in adults and children. *Journal of sleep research.* 2021:e13387.
22. Sivam S, Chamula K, Swieca J, Frenkel S, Saini B. Narcolepsy management in Australia: time to wake up. *The Medical Journal of Australia.* 2021;215(2):62-63. e61.
23. Miserandino C. The spoon theory. In: *Beginning with Disability.* Routledge; 2017:174-178.
24. Quinn DM, Williams MK, Quintana F, et al. Examining effects of anticipated stigma, centrality, salience, internalization, and outness on psychological distress for people with concealable stigmatized identities. *PloS one.* 2014;9(5):e96977.
25. Derickson A. *Dangerously Sleepy: Overworked Americans and the Cult of Manly Wakefulness.* University of Pennsylvania Press, Incorporated; 2014.
26. Alger SE, Brager AJ, Capaldi VF. Challenging the stigma of workplace napping. *Sleep.* 2019;42(8):zsz097.
27. McEwen BS. Central effects of stress hormones in health and disease: Understanding the protective and damaging effects of stress and stress mediators. *European journal of pharmacology.* 2008;583(2-3):174-185.
28. Lishman WA. Organic psychiatry. *The psychological consequences of cerebral disorder.* 1987;127.
29. Morse AM, Sanjeev K. Narcolepsy and psychiatric disorders: comorbidities or shared pathophysiology? *Medical Sciences.* 2018;6(1):16.
30. Barateau L, Lopez R, Chenini S, et al. Depression and suicidal thoughts in untreated and treated narcolepsy: systematic analysis. *Neurology.* 2020;95(20):e2755-e2768.
31. Nigam M, Hippolyte A, Dodet P, et al. Sleeping through a pandemic: impact of COVID-19–related restrictions on narcolepsy and idiopathic hypersomnia. *Journal of Clinical Sleep Medicine.* 2022;18(1):255-263.