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Medical specialty visits and diagnoses received by Saudi patients prior to a diagnosis of narcolepsy

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Abstract

Purpose Narcolepsy is an uncommon neurological disorder characterized by excessive daytime sleepiness (EDS) and multiple other symptoms. Due to the under-recognition of narcolepsy symptoms, patients are often misdiagnosed. This study aimed to assess the types of specialties visited and the diagnoses received by Saudi patients prior to their narcolepsy diagnosis.

Methods The study included 55 consecutive patients with type-1 and type-2 narcolepsy who attended the narcolepsy clinic between August 2017 and December 2017. Narcolepsy was diagnosed according to the International Classification of Sleep Disorders–third edition criteria. We evaluated sociodemographic data, the specialties visited, and diagnoses and treatments received prior to visiting a sleep specialist.

Results The mean diagnostic delay was 9.1 ± 8.4 years (1–43 years). Multiple linear regression analysis identified early onset as the only predictor of a delayed diagnosis (β coefficient = − 0.262, $p = 0.03$). EDS was the main symptom that prompted patients to seek medical consultation, and only one patient had been (1.8%) referred with the diagnosis of narcolepsy. In the study group, 82% of the patients were misdiagnosed with a mental or neurological disorder or were thought to be afflicted by "envy," "evil eye," or "black magic" before receiving a correct diagnosis. No significant differences were detected between patients with narcolepsy type-1 and narcolepsy type-2.

Conclusions Delays in diagnosing narcolepsy remain a major problem for Saudi patients with this disorder. We found that Saudi patients with narcolepsy had visited several medical specialists and faith healers and were misdiagnosed prior to visiting a sleep specialist.

Keywords Depression, neurological disorder · Misdiagnosis · Hypersomnia · Diagnostic delay · Cataplexy

Introduction

Narcolepsy is an uncommon neurological disorder characterized by a pentad of symptoms that include excessive daytime sleepiness (EDS), hypnagogic hallucinations, cataplexy, sleep paralysis, and disturbed nocturnal sleep [1]. It is classified into two types: type-1, associated with cataplexy; and type-2, without cataplexy [1]. Narcolepsy affects approximately 25 to 50 out of 100,000 individuals [2]. The estimated prevalence in Saudi Arabia is 40 in 100,000 (0.04%) [3]. As it is an uncommon disorder, there is often a lack of awareness among both medical professionals and the general public, which may ultimately lead to delays in making a correct diagnosis and proving proper treatment [4]. Delayed narcolepsy diagnoses are consistently reported across countries, as patients frequently receive a correct diagnosis years after the onset of symptoms [4]. A previous study in Saudi Arabia reported that the interval between the onset of narcolepsy symptoms and diagnosis was more than 8 years [5]. Studies conducted in several Western countries over the last decade have demonstrated a trend towards a more timely diagnosis [4, 6]. Nevertheless, these persistent delays in making the correct diagnosis reflect an under-recognition of narcolepsy within the medical community. Previous studies suggest that the under-recognition of narcolepsy symptoms is the main reason for a delayed diagnosis [4, 7–9]. A limited number of studies have assessed prior diagnoses received by narcolepsy patients and none have been

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conducted in Saudi Arabia or other Arab countries [6, 10–12]. It has been reported that patients with narcolepsy received several mental and neurological diagnoses before receiving a correct diagnosis [6, 10]. Moreover, racial and cultural differences in narcolepsy presentation have been reported, and this variability in symptoms may result in receiving alternative diagnoses [5, 13]. We hypothesized that diagnostic delay of Saudi narcolepsy patients has not improved, and that these patients visited several specialties and received several diagnoses before receiving the correct diagnosis. Therefore, the objective of this study was to assess the specialties visited and the diagnoses received by Saudi narcolepsy patients prior to receiving a correct diagnosis.

Methods

Subjects

This study was approved by the Ethics Committee of the College of Medicine, King Saud University. Informed written consent was obtained from all participants. The study included 55 consecutive patients attending a narcolepsy clinic in the University Sleep Disorders Center (USDC) at King Saud University Medical City between August 2017 and December 2017. The USDC is a national tertiary-care sleep disorders center that accepts referrals of all age groups from across Saudi Arabia and provides immediate access to all patients with suspected narcolepsy. Furthermore, all Saudis have free access to government-funded healthcare services. One of the authors (ASB) evaluated all the cases in this study.

The diagnosis of narcolepsy was based on the International Classifications of Sleep Disorders–third edition (ICSD-3) [1]. All patients underwent full polysomnography followed by a multiple sleep latency test (MSLT) [1]. In addition to irresistible attacks of sleep, narcolepsy type-1 was diagnosed by a mean latency of < 8 min on the MSLT with evidence of sleep-onset rapid eye movement periods (SOREMPs) and clear cataplexy ("more than one episode of generally brief (< 2 min), usually bilaterally symmetrical, sudden loss of muscle tone with retained consciousness"). Narcolepsy type-2 was diagnosed by a mean latency of < 8 min on the MSLT and two SOREMPs (or one SOREMP on PSG and one or more on MSLT), but without cataplexy [1]. Other causes of excessive daytime sleepiness were excluded.

Data pertaining to the symptoms at onset, age of disease onset, and age at the time of diagnosis were obtained from the USDC database.

Questionnaire

The questionnaire was adapted from previous studies [6, 10], and consisted of 53 items designed to collect the following data: sociodemographic data, reasons for the delay in diagnosis, specialties consulted, and the diagnoses and treatments received before visiting a sleep specialist. Daytime sleepiness was assessed using a validated Arabic version of the Epworth sleepiness scale (ESS) [14, 15]. Most items describe situations or contain statements. Other items are questions and the patients had a choice between a few different answers, with the possibility to mark multiple statements or to make additional comments. Other questions inquired about diseases and medical conditions, comparing the situations before diagnosis and at the moment of study conduction [6]. The questionnaire was piloted on five patients to evaluate the feasibility of data collection, reliability and accuracy of data collection tools, and estimation of timing for data collection. Data collection was performed via face-to-face interview. On average, 12 min were needed to complete the questionnaire. Adjustments were applied to the questionnaires based on the feedback received from the participants.

Statistical analysis

Data were analyzed using the Statistical Package for Social Sciences (SPSS, version 22). Group data were described as mean ± standard deviation or numbers and percentages. Comparisons between narcolepsy type-1 and narcolepsy type-2 were performed using Student's t test for normally distributed variables; otherwise, the Mann-Whitney U test was used. For nominal data, chi-square tests were applied. To identify predictors of a prolonged delay in making the correct diagnosis, stepwise multiple linear regression analysis was performed including the following independent variables: age of disease onset, body mass index, sex, the number of symptoms at the time of diagnosis, household income, educational level, and a history of misdiagnosis with a psychiatric or neurologic disease. A p value < 0.05 was considered statistically significant.

Results

A total of 55 patients (males 84%) were included in the study. The mean body mass index was 31 ± 8 kg/m^2 and the mean ESS score was 18.5 ± 3.0. Slightly more than half of the patients (58.2%) had obtained a bachelor's degree. The demographic data are shown in (Table 1). The mean diagnostic delay was 9.1 ± 8.4 years. The diagnostic delay ranged between 1 and 43 years with a median of 7.5 years. Multiple linear regression analysis identified early age of onset as the only predictor of a prolonged delay in diagnosis (β coefficient = − 0.262, CI = − 0.501–− 0.024, and p = 0.03).

Eighty-two percent of our sample group were misdiagnosed with another mental or neurological disorder or were thought to be afflicted by "envy"/"evil eye" or "black

Table 1 Demographic, clinical, and general characteristics

Variable	*Total (n = 55)*	*Mean ± SD/n (%)/median (25th–75th percentile)*
Body mass index (kg/m^2)		31 ± 8 (25.7–35.6)
Sex (male)		46 (84%)
Age at diagnosis		30.9 ± 10.6
Age at onset		21.7 ± 9.3
Time between onset of symptoms and diagnosis (years)		9.1 ± 8.4
Epworth sleepiness scale		18.5 ± 3.0
Narcolepsy symptoms at time of onset		
Irresistible attacks of sleep (excessive daytime sleepiness)		55 (100)
Hypnagogic		33 (62.3)
Hypnopompic		10 (18.9)
Sleep paralysis		32 (60.4)
Cataplexy		29 (53.7)
Interrupted sleep		25 (47.2)
Which symptom pushed you to seek the first medical visit?		
Excessive daytime sleepiness		45 (81.8)
Cataplexy		9 (16.4)
Hypnogogic hallucination		2 (3.6)
Sleep paralysis		12 (21.8)
Educational level		
Primary level		1 (1.8)
Intermediate level		3 (5.5)
Secondary level		18 (32.7)
Bachelor's degree		32 (58.2)
PhD		1 (1.8)
Employment		
Part-time		3 (5.8)
Full-time		35 (67.3)
Unemployed		14 (26.9)

magic" prior to being correctly diagnosed with narcolepsy. EDS was the main symptom that prompted patients to seek medical consultation. Only one patient (1.8%) had been referred to a sleep specialist with the diagnosis of narcolepsy, which was done by a pulmonary specialist. Of the remaining cases, the patients were referred to a sleep specialist to assess their daytime sleepiness or disturbed nocturnal sleep.

Table 2 presents the characteristics of the patients' journey before visiting a sleep specialist and receiving the correct diagnosis. The majority of patients (54.5%), when asked, claimed not to know the cause behind the delay in their diagnosis, 27.3% felt that the treating doctors' knowledge about sleep disorders were inadequate, 21.8% indicated that the treating doctor did not refer them to a sleep specialist, and 10.9% blamed themselves for a delay in seeking medical advice.

Prior to receiving the correct diagnosis of narcolepsy, the study showed that the patients had visited the following providers: a spiritual (faith) healer ($n = 31$, 56.4%), a neurologist ($n = 19$, 34.5%), a psychiatrist ($n = 18$, 33.3%), an otolaryngologist ($n = 4$, 7.3%), an emergency department ($n = 3$, 5.5%), and an internist ($n = 3$, 5.5%). Approximately 82% of the patients were misdiagnosed with a mental, neurological, or spiritual problem before receiving a correct diagnosis. Patients were told that their symptoms were a result of "envy"/"evil eye" ($n = 22$, 40%), a psychiatric disorder ($n = 11$, 20%), a neurological disease ($n = 6$, 11%), or black magic ($n = 6$, 11%). Around 13% ($n = 7$) indicated that they used herbal medications before being diagnosed with narcolepsy, and 20% ($n = 11$) were prescribed psychiatric medications.

Apart from a higher rate of envy/evil eye diagnosis, no significant differences were detected between patients with narcolepsy types 1 and 2 in terms of diagnostic delay or other parameters (Table 3).

Discussion

Our findings show that there is still a significant delay in correctly diagnosing narcolepsy in Saudi patients. The majority of patients with narcolepsy visited with neurology, psychiatry, and otolaryngology specialists or faith (spiritual) healers, and received different diagnoses before visiting with a sleep specialist. Additionally, during those visits, they were misdiagnosed with having psychiatric and neurological disorders or were thought to be afflicted by "evil eye" or black magic. Our results concur with those from previous studies, which show that patients with narcolepsy are often misdiagnosed with a variety of mental and neurological disorders [6, 7, 10, 11, 16, 17]. Symptoms of several neuropsychiatric disorders overlap with those of narcolepsy, which frequently results a misdiagnosis [18].

A previous study in Canada demonstrated that neurologists were the least likely to misdiagnose narcolepsy compared to other specialists; they accurately diagnosed narcolepsy in 55% of cases presented to them [10]. However, this was not the case in our study, as none of the patients seen by a neurologist was correctly diagnosed.

The paroxysmal attacks of cataplexy seen in narcolepsy may be confused with sudden drop attacks seen in some neurological disorders. Reports have shown that in some cases, drop attacks suffered by patients diagnosed with syncope and epilepsy may mimic cataplexy [17, 19, 20]. Such misdiagnoses result in delays with reaching the correct diagnosis and providing proper treatment. For example, treatment with anticonvulsants has been reported in narcolepsy patients who were misdiagnosed with epilepsy [17].

Psychiatry is the second most frequently visited specialty by patients prior to being diagnosed with narcolepsy. One

Table 2 Characteristics of the patients' journey before visiting a sleep specialist

Variable	*Total (n = 55)*	*n (%)*
In your opinion, what is the cause of delayed diagnosis of narcolepsy?		
I do not know		30 (54.5)
The treating doctor did not refer me to a sleep specialist		12 (21.8)
I did not know that there is a sleep medicine specialty		15 (27.3)
Doctors' knowledge about sleep disorders is inadequate		15 (27.3)
Delay in seeking medical advice		6 (10.9)
Other causes		6 (10.9)
List the specialties you visited before visiting a sleep specialist		
Psychiatry		18 (33.3)
Neurology		19 (34.5)
Emergency department		3 (5.5)
Pediatrics		0 (0)
Other departments		14 (25.5)
Pulmonary medicine		2 (3.6)
Otolaryngology		4 (7.3)
Internal medicine		3 (5.5)
Have you previously visited a faith (spiritual) healer?		31 (56.4)
Were you previously misdiagnosed with any of these disorders before your narcolepsy diagnosis?		
Psychiatric disorder		11 (20)
Neurological disorder		6 (10.9)
Black magic		6 (10.9)
"Envy" and "evil eye"		22 (40)
Did you ever attend any psychotherapy or cognitive behavioral therapy session before your narcolepsy diagnosis? (Yes)		14 (25.5)
Did you use any psychiatric medication? (Yes)		11 (20)
Do you use any herbal medication? (Yes)		7 (12.7)

third of our study sample visited psychiatry clinics, and a proportion of them were prescribed psychiatric drugs and psychotherapy. The presence of hallucinations and other psychotic symptoms in narcolepsy patients have been reported as potential factors responsible for misdiagnoses with schizophrenia and other psychiatric disorders [8, 9, 21, 22]. A previous Canadian study examined all visits made by narcolepsy patients to psychiatrists before receiving a proper diagnosis and reported that only 11% of psychiatrists made the correct diagnosis [10]. Furthermore, several studies have demonstrated an association between narcolepsy and other psychiatric disorders [23]. Therefore, narcolepsy patients may have comorbid psychiatric disorders [18]; however, attributing narcolepsy symptoms to a psychiatric disorder and not detecting the coexistence of narcolepsy results in a significant delay in diagnosis.

Surprisingly, otolaryngology was frequently visited by Saudi narcolepsy patients before receiving a correct diagnosis. This has not been previously reported. Because most otolaryngologists in Saudi Arabia are interested in the surgical treatment of obstructive sleep apnea, this may mislead patients to think that they are specialized in sleep medicine [24].

Interestingly, 40% and 11% of narcolepsy patients were diagnosed by faith (folk or spiritual) healers as being afflicted by "envy/evil eye" and black magic, respectively. "Evil eye" is a common belief in several cultures and religions, in which people believe that a glance may have the power to cause injury and diseases in those to whom it falls [25, 26]. In Saudi Arabia, it is not uncommon for faith healers to attribute mental and psychiatric disorders to "envy/evil eye" and black magic [27]. Faith healers consider supernatural etiology for mental disorders [27, 28]. In general, beliefs about Jinn, black magic, and the "evil eye" and their effects on the physical or mental health of humans are not uncommon in Saudi Arabia and other Muslim cultures at large [27, 29–34]. A previous study that examined community attitudes towards auditory hallucinations in Saudi Arabia and the United Kingdom (UK) indicated that those living in Saudi Arabia were most likely to believe that the hallucinations are caused by Satan (evil) or black magic, whereas the UK study population was more likely to consider schizophrenia or brain damage [34]. While the Saudi study group considered faith and religious healers to be most helpful, the UK study group opted for medication and psychological therapies. Interestingly, these cultural beliefs regarding disease etiology and treatment were not influenced by educational level [34].

The current findings apply to Muslims in different cultures including those living in Western societies. A study that was conducted in the UK assessed Muslims' beliefs about Jinn, black magic, and the evil eye showed that the majority of the sample believed in the existence of jinn, black magic, and the evil eye [30]. Additionally, approximately, half of the sample stated that these could cause physical and mental health problems and that these problems should be treated by both doctors and religious (faith) healers [30]. Another recent study in the Netherlands that recruited Muslim patients attending a transcultural outpatient clinic specialized reported that attributing mental health symptoms to jinn was much more common among Muslim patients than previously assumed [29]. The above emphasizes the need for proper knowledge of Muslim explanatory models of disease and for the use of culturally sensitive interviewing techniques.

These findings suggest that clinicians should be open to the religious and cultural viewpoints of their patients' problems and to be prepared to collaborate with faith healers to avoid an unnecessary delay in diagnosis and begin proper treatment [30]. Nevertheless, further research is needed to assess the effect of these cultural and religious beliefs in the delay in diagnosing mental and physical illnesses.

Table 3 Comparison between narcolepsy type-1 and type-2 patients

Variable	*Total (n = 55)*	*Mean ± SD/n (%)*		*p value*
		NT1 (29)	*NT2 (25)*	
Body mass index (kg/m^2)		31.8 ± 9.2	30.5 ± 6.3	0.910
Age of disease diagnosis		29.2 ± 11.3	32.8 ± 9.5	0.090
Age at onset		20 ± 8.2	23.7 ± 10.3	0.301
Time between onset of symptoms and diagnosis (years)		9.2 ± 10.2	9 ± 5.9	0.229
Sex (men)		24 (82.8)	21 (84)	1.000
List the department you visited before visiting a sleep specialist				
Psychiatric department		10 (35.7)	7 (28)	0.548
Neurology department		10 (34.5)	9 (36)	0.907
Emergency department		3 (10.3)	0 (0)	0.240
Pediatric department		0 (0)	0 (0)	–
Other departments		8 (27.6)	6 (24)	0.764
Pulmonary medicine		1 (3.4)	1 (4)	0.956
Otolaryngology		2 (6.9)	2 (8)	0.95
Internal medicine		2 (6.9)	1 (4)	0.67
Have you previously visited a faith (or spiritual) healer? (Yes)		18 (62.1)	13 (52)	0.456
Were you previously misdiagnosed with any of these disorders before your narcolepsy diagnosis?				
Psychiatric disorder		5 (17.2)	6 (24)	0.539
Neurological disorder		4 (13.8)	2 (8)	0.675
Black magic		2 (6.9)	4 (16)	0.399
"Envy"/"evil eye"		16 (55.2)	6 (24)	0.020
Do you use any herbal medications? (Yes)		4 (13.8)	3 (12)	1.000
Did you ever attend any psychotherapy or cognitive behavioral therapy session before diagnosed you with narcolepsy? (Yes)		9 (31)	5 (20)	0.356
Did you use any psychiatric medication? (Yes)		8 (27.6)	3 (12)	0.156

An older study conducted in Saudi Arabia demonstrated that among 47 narcolepsy patients who were referred to the sleep disorders clinic, only three patients were correctly diagnosed by their referring doctor [5]. In the current study, only one patient was correctly diagnosed by the referring doctor. Additionally, in the present study, the time gap between the onset of symptoms and narcolepsy diagnosis (9.1 years) had not improved when compared to a previous Saudi study (conducted 13 years ago), which reported a delay of 8.4 years. This indicates that Saudi physicians' knowledge and awareness of narcolepsy has not improved. Previous studies reported several predictors of delayed narcolepsy diagnosis, such as female sex, body mass index, and age of disease onset [6, 35, 36]. On the contrary, the best predictor was the presence of cataplexy at presentation to medical care services [6, 35, 36]. In our study, early onset was the only predictor of diagnostic delay. This concurs with the results of a recent study from Switzerland, which identified early age of disease onset as the strongest predictor of prolonged diagnostic delay [6]. This finding could be related to difficulties in interpreting these symptoms in children. The presence of cataplexy at the time of symptom onset was not a predictor of shorter diagnostic delay in the current study. These findings concur with those of an earlier Saudi study [5] and with a recent Austrian study of 100 narcolepsy patients [37]. The discrepancy between these various studies could be attributed to differences in the studied populations.

The under-recognition and delay in diagnosing narcolepsy is unlikely to be secondary to reduced access to the medical care, as all Saudi citizens have free access to primary care services in their communities; additionally, the University Sleep Disorders Center at King Saud University provides immediate access to all Saudi patients with suspected narcolepsy [38]. Even in developed countries, the delay in diagnosis does not seem to be related to accessibility to healthcare services. A previous Canadian case-control study demonstrated that narcolepsy patients have double the number of doctors' visits compared to controls [10]. Thus, it is likely that the misdiagnosis of narcolepsy reflects a lack of recognition of narcolepsy symptoms. A recent national survey of selected Saudi medical schools used a standardized tool to assess medical students' sleep medicine knowledge and revealed that only 4.6% of students had sufficient knowledge [39]. Similarly, the exposure of postgraduate students to sleep disorders during residency training is limited [40]. Previous surveys that aimed to assess primary care physicians' knowledge in Saudi Arabia revealed that their sleep medicine knowledge was limited [39, 41–43].

These findings suggest that insufficient education in sleep medicine, at both the undergraduate and postgraduate levels, has resulted in a medical community with limited knowledge of sleep disorders, which may affect the ability to properly diagnose them.

In a recent survey that evaluated the understanding and perceptions of narcolepsy among a sample of physicians, including sleep specialists in the US, only 39% and 63% were able to identify cataplexy and excessive daytime sleepiness, respectively, as the two major symptoms of narcolepsy [44]. Moreover, only one-fifth of sleep specialists were able to recognize the five symptoms of narcolepsy [44].

Another interesting finding of this study is that only 16% of the patients were women, which is not similar to data reported in other countries [2]. The current results conform to previously published data in Saudi Arabia showing that men represent 80% of narcolepsy patients [38]. A possible explanation for this difference could be the conservative nature of Saudi society where women could be less forthcoming with their symptoms, preferring self-medicate, or because they are less affected in their daily lives compared to men [45, 46].

The current study has some limitations. In general, questionnaires are prone to recall bias and some statements may be unclear. Nevertheless, our results are in agreement with those of previously published data in other countries, which may reflect the reliability of the current data. Another limitation is that there is no electronic national database available for all patients, making it impossible to track all physician visits, associated diagnoses, and prescribed medications. Nonetheless, this is the first study to address diagnoses received by narcolepsy patients prior to visiting a sleep specialist in Saudi Arabia, and to report new data in a country that is culturally and ethnically different from Western countries.

In conclusion, delays in the diagnosis of narcolepsy remain a major problem for Saudi patients with this disorder. Patients with narcolepsy appeared to visit several medical specialists and faith healers and were frequently misdiagnosed prior to consulting with a sleep specialist. Intensive educational programs on sleep disorders, particularly those that target both the medical community and the general public, are unquestionably needed. Local training programs for the specialties frequently visited by patients with sleep disorders—such as psychiatry, neurology, otolaryngology, and primary care—should have both theoretical and practical training in sleep medicine. Thus, increased education and awareness may facilitate the early detection of sleep disorders, the provision of proper treatment, and the prevention of complications. Additionally, future studies should assess the psychosocial consequences of the diagnostic delay.

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Compliance with ethical standards

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Informed consent Informed consent was obtained from all participants.

Conflict of interest The authors declare that they have no conflict of interest.

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