Narcolepsy is a lifelong neurologic disease for which no cure has been identified. Narcolepsy is characterized by excessive daytime sleepiness and signs and symptoms of sleep/wake disarray such as cataplexy, rapid transitions into rapid eye movement sleep, sleep paralysis, hypnagogic hallucinations, and disrupted nocturnal sleep. The onset of narcolepsy almost always occurs after 5 years of age and typically between ages 15 and 25 years. Narcolepsy is associated with high healthcare resource utilization and substantial socioeconomic burden related to reductions in quality of life, unemployment, and reduced productivity. A delay of approximately 10–15 years between the onset of narcolepsy symptoms and receiving a narcolepsy diagnosis has been reported in the literature, this suggests that there are challenges throughout the journey to a narcolepsy diagnosis. — Delays in receiving a diagnosis of narcolepsy might occur for a variety of reasons, such as the presence of comorbid conditions and the misdiagnosis of narcolepsy as another sleep or neurological disorder.

Objective

To conduct a physician survey and chart review in order to characterize patients’ journeys on their way to a diagnosis of narcolepsy.

Methods

Online quantitative patient chart survey fielded among physicians from May 23 to June 18, 2012

Physicians with neurology, pulmonology, or psychiatry as a primary specialty were recruited from a national representative database (Cognitart Analytics opt-in panel; N=4457 physicians invited to participate) or from a company-generated list of healthcare providers who are likely to treat narcolepsy patients (N=4681 physicians invited)

To qualify for the study, physicians had to meet the following criteria:

— Have between 2 years and 30 years of clinical experience
— Spend more than 75% of their time in patient care
— Treat at least 30 patients per month, including at least 5 narcolepsy patients

Physicians were invited via emails and postal invitations, and the survey/chart review was conducted online. Each physician was asked to complete up to 6 questionnaires using the charts of patients they treated for narcolepsy within the last 6 months. Each patient chart questionnaire took approximately 10 minutes to complete.

All physicians provided informed consent prior to their participation in the study and were compensated for each patient chart questionnaire that they completed.

Results

A total of 252 patient charts were collected from 77 physicians (Table 1).

Table 1. Participating Physicians by Specialty

<table>
<thead>
<tr>
<th>Specialty</th>
<th>No. of Physicians (No. of Patients)</th>
<th>n=134</th>
<th>n=138</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neurology</td>
<td>18 (55)</td>
<td>19 (54)</td>
<td></td>
</tr>
<tr>
<td>Pulmonology</td>
<td>17 (80)</td>
<td>5 (56)</td>
<td></td>
</tr>
<tr>
<td>Psychiatry</td>
<td>5 (17)</td>
<td>12 (88)</td>
<td></td>
</tr>
<tr>
<td>Other</td>
<td>1 (0)</td>
<td>0</td>
<td></td>
</tr>
</tbody>
</table>

BSCM, board certified sleep medicine

Survey respondents 72%

Table 1: Participating Physicians by Specialty

- The patient sample was predominantly male (65%), with a median age of 38 years (Figure 1)
- At least half of the patients were overweight or obese (median body mass index 25.8 kg/m²).
- Approximately two-thirds of the patients had a comorbid condition
- Psychiatric and metabolic comorbidities were common (nearly one-third of patients had comorbid depression).
- Only 3% of patients in this sample were uninsured.

Figure 1. Patient Demographics and Characteristics

**METHODS**

1. **Delays in receiving a diagnosis of narcolepsy**

— Treat at least 30 patients per month, including at least 5 patients with narcolepsy
— Spend more than 75% of their time in patient care
— Have at least 1 year of experience with narcolepsy
— See at least 30 patients per month, including at least 5 patients with narcolepsy

**PATIENT DEMOGRAPHICS & CHARACTERISTICS**

— The respondent physician made the diagnosis of narcolepsy in 72% of cases in the 27th Annual Meeting of the BSCM, board certified in sleep medicine.
— Polysomnography (PSG), multiple sleep latency test (MSLT), and Epworth Sleepiness Scale (ESS) in 65%, 70%, and 62% of patients, respectively.
— Cerebrospinal fluid testing for hypocretin-1 was only performed in 2% of patients.
— The majority of patients were characterized by short sleep latency (<5 minutes), sleep latency on the MSLT, and severe sleepiness (ESS score >10).
— The mean sleep latency on the MSLT was 4.2 minutes and the mean ESS was 17.0.
— On the MSLT, 91% of patients had ≥2 periods of sleep-onset rapid eye movement

Figure 2. Getting to the Right Place—the Referral Process

**GETTING THE RIGHT TESTS—THE DIAGNOSTIC PROCESS**

— Excessive daytime sleepiness was the most common symptom at initial presentation (90% of patients) (Figure 3).
— Over 40% of patients reported trouble staying awake and functioning during the day; one-quarter reported difficulties with activities of daily living.
— The majority of patients (85%) were rated as having moderate to severe symptoms at their initial visit, with more than one-third having severe symptoms.
— In 44% of cases the patient’s family/spouse helped confirm the symptoms.
— The most commonly performed assessments were polysomnography (PSG), multiple sleep latency test (MSLT), and Epworth Sleepiness Scale (ESS), in 65%, 70%, and 62% of patients, respectively.
— Cerebrospinal fluid testing for hypocretin-1 was only performed in 2% of patients.
— The majority of patients were characterized by short sleep latency (<5 minutes), sleep latency on the MSLT, and severe sleepiness (ESS score >10).
— The mean sleep latency on the MSLT was 4.2 minutes and the mean ESS was 17.0.
— On the MSLT, 91% of patients had ≥2 periods of sleep-onset rapid eye movement

Figure 3. Getting the Right Tests—the Diagnostic Process

**SUMMARY OF THE PATIENTS’ JOURNEYS**

— Narcolepsy was misdiagnosed in 41% of cases, with 40% of patients being misdiagnosed as other sleep or neurological disorders.
— Narcolepsy was misdiagnosed in 38% of cases, with 39% of patients being misdiagnosed as other sleep or neurological disorders.
— Narcolepsy was misdiagnosed in 29% of cases, with 30% of patients being misdiagnosed as other sleep or neurological disorders.
— Narcolepsy was misdiagnosed in 19% of cases, with 20% of patients being misdiagnosed as other sleep or neurological disorders.
— Narcolepsy was misdiagnosed in 9% of cases, with 10% of patients being misdiagnosed as other sleep or neurological disorders.

Figure 4. Summary of the Patients’ Journeys

**REFERENCES**


**DISCLOSURES:**

Drs Carter and Abeo, and Ms Kim are shareholders of Jazz Pharmaceuticals plc and employees of Jazz Pharmaceuticals Inc. who, in the course of this employment, have received stock options exercisable for, and other stock awards of, ordinary shares of Jazz Pharmaceuticals plc.

**Support:** This research was funded by Jazz Pharmaceuticals, Inc. The authors would like to thank Gregg Checani, MD, François Di Trapani, PharmD, and Trudy Vanhove, MD, PhD, MBA, from Jazz Pharmaceuticals, Inc. for our review of the poster for accuracy. E Jay Bienen, PhD, of The Curry Rockefeller Group, LLC (CRG), provided editorial assistance in formatting, proofreading, copy editing, and fact checking. Jazz Pharmaceuticals, Inc. provided funding to CRG for support in writing and editing this poster.