Treatment patterns for Charcot-Marie-Tooth disease in the UK and US: insights from a digital real-world observational study

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BACKGROUND
Charcot-Marie-Tooth disease (CMT) is a hereditary motor and sensory neuropathy that affects the peripheral nervous system, leading to muscle atrophy and impaired sensitivity to touch, vibration, heat and pain. CMT compromises patient lifestyles, everyday activities, and career and family choices. CMT is rare, and there has been little research into its impact on patients’ lives. The collection of real-world data, direct from patients, may therefore provide valuable insights.

OBJECTIVES
The objective of this analysis was to examine patient-reported treatment patterns for CMT in UK and US real-world practice.

METHODS
Adults with CMT were recruited to an ongoing two-year international observational study exploring the real-world burden of the condition. Data were collected via CMT&Me, a ‘bring your own device’ smartphone app, through which participants were asked to provide data on demographic, CMT management-related and quality-of-life variables. This updated interim analysis (data cut 30 May 2019, approximately seven months into the study) examined participants’ responses to in-app surveys about demographic characteristics and CMT treatment patterns, including whether they had ever received the following treatments as part of their CMT care:
- Rehabilitation therapies
- Medications
- Aids and orthoses
- Surgical procedures.

RESULTS

Demographics
Characteristics of participants who responded to demographic profile questions are presented in Table 1. The proportions of respondents from the UK and US were similar. Almost two thirds of respondents were women. The proportions of respondents aged <50 and ≥50 were similar. The most common CMT subtype was CMT1A, followed by CMT2 and Unknown. Note: as most questions in the app were optional, respondents to the demographics questions were not necessarily the same participants who responded to questions about CMT treatments.

<table>
<thead>
<tr>
<th>Demographics</th>
<th>Value</th>
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<tbody>
<tr>
<td>Respondents (n)</td>
<td>666</td>
</tr>
<tr>
<td>Country of residence (%), n</td>
<td>330 (49.0) 336 (50.0)</td>
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</tbody>
</table>

A slightly higher mean proportion of women than men reported use of rehabilitation therapies (24% vs 17%, respectively). Slightly higher use in women than men was observed across the majority of therapy types. Similar mean proportions of respondents aged <50 and ≥50 reported receiving rehabilitation therapies (24% vs 22%, respectively). The mean proportion of women reporting use of aids was similar to the proportion of men (18% vs 17%, respectively). Across most types of aids a slightly higher proportion of women than men reported use. Overall, mean use of aids was similar in respondents aged ≥50 and <50 (17% vs 19%, respectively). However, there was a particularly high proportion of walking stick users in the older age group.

Medications
The majority of respondents had received medication for their CMT, the most common classes being non-opioid analgesics and antidepressants (Figure 1). On average, respondents had received 1.9 different classes of medication. The mean proportion of recipients across all medication types was slightly greater in the US than in the UK (20% vs 16%, respectively). The CMT subtypes with the highest mean proportions of respondents reporting medication use were HNPP and CMT4 (both 23%), followed by CMT2 (21%). However, these results should be interpreted with caution given very small sample sizes. A slightly higher mean proportion of women than men reported use of medication (20% vs 13%, respectively). This trend held true across all medication classes.

Aids and orthoses
Almost 90% of respondents had used aids or orthoses for their CMT, the most common types being ankle/foot braces, walking sticks, and insoles (Figure 2). On average, respondents had used 2.5 different types of aid. Similar mean proportions of UK and US respondents reported using aids and orthoses, with the average proportions of users across all aid types being 19% and 17%, respectively. The highest mean proportional use of aids was in respondents with CMT5 (21%), followed by Unknown CMT subtypes, CMT2, and CMT3 (all 19%).

Surgical procedures
Around half of respondents had undergone a surgical procedure for their CMT, the most common being ostectomy, hammertoe correction, and planter fascia release (Figure 3). Among all respondents, the mean number of surgical procedures received was 1.2.

The mean proportion of surgery recipients across all procedure types was 9% in both the UK and US. This similarity was consistent across all procedure types.

The CMT subtypes reporting highest mean proportional receipt of surgical procedures were CMT3 (15%), CMT4 (12%), and CMT1A (11%). However, the results for CMT3 and CMT4 in particular should be interpreted with caution given the very small sample sizes.

Similar mean proportions of women and men had undergone surgery (10% vs 8%, respectively). Similar mean proportions of respondents aged <50 and ≥50 reported receiving surgery (10% vs 9%). This similarity was particularly high proportion of walking stick users in the older age group. Similar mean proportions of women and men had undergone surgical procedures (10% vs 9%). This similarity was particularly high proportion of walking stick users in the older age group. Similar mean proportions of people in the younger age group had received hip or knee surgery.

REFERENCES
1. Owens GM, J Manag Care Pharm. 2008;14(3;suppl S):S2–6

CONCLUSIONS
The management of CMT in the UK and US is multifaceted, involving the use of physical and surgical therapies, as well as multiple medications, aids, and orthoses. This ongoing registry will provide further real-world insights into the treatment of CMT to enable gaps in care to be identified and addressed.

Figure 1: Rehabilitation therapies

Figure 2: Medications

Figure 3: Aids and orthoses

Figure 4: Surgical procedures