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 1: Demographics

Parameter	Value
Respondents (n)	666
Country of residence (n,%)	
UK	330 (49.5)
US	336 (50.5)
Sex (n,%)	
Female	455 (68.3)
Male	211 (31.7)
Age (years)	
Mean	48.1
Standard deviation	14.7
Median	49.0
Range	18–82
Age <50 (n,%)	341 (51.2)
Age ≥50 (n,%)	325 (48.8)
CMT subtype (n,%)	
CMT1A	287 (43.1)
Other subtypes of CMT1	32 (4.8)
CMT2	73 (11.0)
CMT3	5 (0.8)
CMT4	13 (2.0)
CMT5	2 (0.3)
CMTX	37 (5.6)
Unknown	62 (9.3)
HNPP	9 (1.4)
Not reported	146 (21.9)

*Six respondents did not report their country of residence.

Rehabilitation therapies

The majority of respondents had received rehabilitation therapy for their CMT, the most common forms being physiotherapy/physical therapy and occupational therapy (Figure 1). On average, respondents had received 1.6 different types of physical therapy.

The mean proportion of recipients across all therapy types was slightly higher in the US than in the UK (26% vs 21%, respectively).

The CMT subtypes with the highest mean proportions of respondents reporting receipt of therapy were HNPP (29%), CMT2 (24%), and CMT1A (23%). However, the results for HNPP in particular should be interpreted with caution given the very small sample size.

A slightly higher mean proportion of women than men reported use of rehabilitation therapies (26% 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).

Medications

The majority of respondents had received medication for their CMT, the most common classes being non-opioid analgesics and antidepressants (Figure 2). 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 CMT3 (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.

Similar mean proportions of respondents aged <50 and ≥50 reported receiving medications (18% vs 17%, respectively).

Aids and orthoses

Almost 90% of respondents had used aids or orthoses for their CMT, the most common types being ankle/leg braces, walking sticks, and insoles (Figure 3). 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%).

The mean proportion of women reporting use of aids was similar to the proportion of men (18% vs 17%, respectively). Across most types of aid 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.

Surgical procedures

Around half of respondents had undergone a surgical procedure for their CMT, the most common being osteotomy, hammertoe correction, and plantar fascia release (Figure 4). 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 generally held true across surgery types; however, there were differences between age groups in certain procedures. Higher proportions of people in the younger age group had received osteotomy, plantar fascia release, and tendon surgery, while a greater proportion of people in the older age group had received hip or knee surgery.

DISCUSSION

Use of rehabilitation therapies, medications, aids and orthoses, and surgical procedures was high among people with CMT.

The majority of respondents reported having used rehabilitation therapies, medications and aids/orthoses to manage their CMT, with around half of respondents also reporting receipt of surgical procedures.

Generally, treatment patterns in the UK and US were similar.

Use of treatments appeared to vary across CMT subtypes. However, given the extremely small sample sizes for certain subtypes this should be interpreted with caution.

Treatment use was generally slightly higher in women than men. This should be interpreted in the context that women in general have been shown to have higher healthcare utilization than men[1-3].

Overall use of treatments was similar between age groups. However, there were a few specific treatments used notably more by older people. These were treatments often used by older members of the general population (walking sticks, knee surgery), so may be evidence of changing general healthcare needs over a lifetime, rather than a reflection of the natural history of CMT.

These results should be interpreted in light of the fact that data were digitally self-reported, which raises challenges in verification.

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.

References

[1]Bertakis KD, J Fam Pract. 2000;49(2):147–52
[2]Owens GM, J Manag Care Pharm. 2008;14(3;suppl S):S2–6
[3]Muller CL, Health Care and Gender. New York: Russell Sage Foundation;1990

Figure 1: Rehabilitation therapies

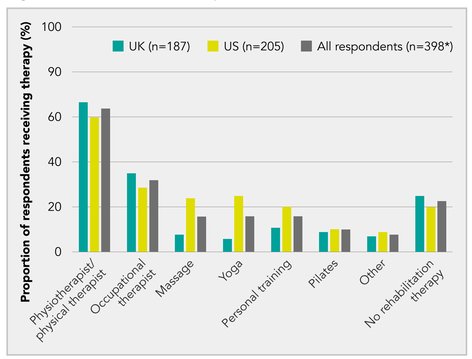

Figure 2: Medications

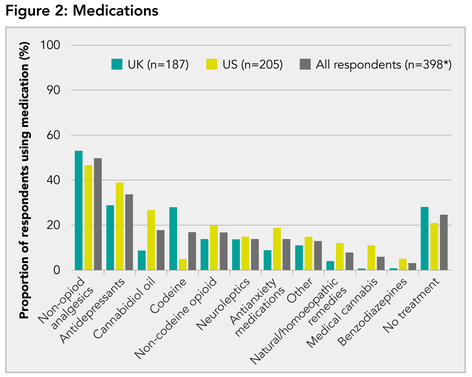

Figure 3: Aids and orthoses

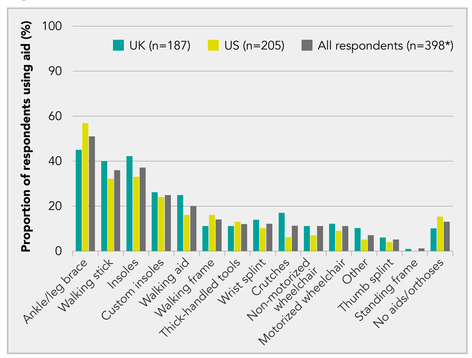

Figure 4: Surgical procedures