Treatment of the Charcot-Marie-Tooth disease population in the United Kingdom and United States: insights from a digital real-world observational study

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Background

Charcot-Marie-Tooth disease (CMT) is a chronic, progressive, motor and sensory neuropathy that affects the peripheral nervous system, leading to distal muscle weakness, muscle atrophy and sensory loss.[1]

CMT compromises patient lifestyles, everyday activities, careers, and family choices.[2] There is currently no curative treatment, meaning that patients are reliant on supportive care.

CMT is rare, and – until recently – there had been little research into its impact on patients' lives. The collection of real-world data, direct from patients, may therefore provide both new and additional valuable insights.

Objectives

The aim of this analysis was to examine patient-reported treatment patterns and care standards for CMT in the UK and US.

Methods

Adults with CMT were recruited to an ongoing two-year international study exploring the real-world burden of the disorder.

Data were collected via CMT&Me, a 'bring your own device' smartphone app, through which participants were asked questions about their CMT and its management.

This interim analysis (data cut August 5, 2019, approximately 9.5 months into the study) examined treatment patterns and care standards reported by UK and US participants.

Results

Disease-management team

Participants were asked about their average annual visit frequency to several professionals considered to be core members of a CMT disease-management team: family doctors, neurologists, and physical/physiotherapists.

Figure 1 presents the proportions of respondents visiting each healthcare professional at least once a year because of their CMT. Family doctors and neurologists were visited annually by most respondents; however, physical/physiotherapists were visited by fewer than half of respondents.

Mean annual visit frequency to each healthcare professional is presented in Figure 2. Physical/physiotherapists were visited most often, followed by family doctors, then neurologists. The high frequency of physical/physiotherapist visits, coupled with the relatively low proportion of respondents visiting annually, indicates that some respondents may have visited particularly frequently. This effect was greater among respondents from the US than from the UK. A slightly higher frequency of visits in the US respondent group was also observed for neurologists.

Figure 1: Proportion of respondents visiting core management team at least once a year

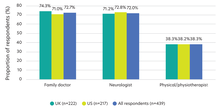


Figure 2: Annual visit frequency to core management team

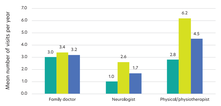
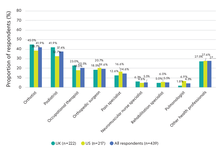


Figure 3 presents the proportions of respondents visiting other types of healthcare professional in the past year because of their CMT. The most commonly visited were orthotists and podiatrists. The proportions of UK and US respondents visiting each healthcare professional were similar.

Figure 3: Proportion of respondents visiting other types of healthcare professionals



CMT supportive care

Participants were asked whether they had ever received various supportive care treatments for their CMT. Figures 4 to 7 present the proportions of respondents who reported receiving rehabilitation therapies, medications, aids/orthoses, and surgical procedures, respectively. Respondents were able to select multiple treatments within each category.

The majority of respondents had received rehabilitation therapy for their CMT at some point, the most common types being physical/physiotherapy, and occupational therapy.

Most respondents had also received medications. The most frequently reported classes were non-opioid analgesics and antidepressants.

A high proportion of respondents reported using orthoses and walking aids, with ankle/leg braces, insoles, and walking sticks being the most commonly reported.

Additionally, around half of respondents had undergone a surgical procedure for their CMT. The most common procedures were osteotomy, hammertoe correction, and plantar fascia release.

Figure 4: Proportion of respondents receiving rehabilitation therapies

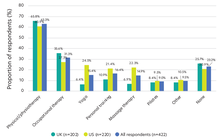


Figure 5: Proportion of respondents receiving medications

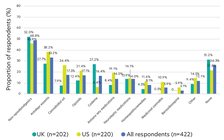


Figure 6: Proportion of respondents using aids/orthoses

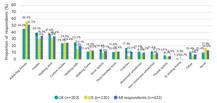
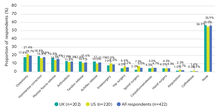


Figure 7: Proportion of respondents receiving surgeries



Discussion

CMT care standards in the UK and US were generally consistent with guidelines[3–8].

As recommended, most participants in this study had at least yearly access to several members of a multidisciplinary disease-management team, including family doctors and neurologists. However, physical/physiotherapists (another core professional within the management team) were visited annually by fewer than half of respondents. This may be reflective of access limitations, indicating that most patients are well-managed so do not regularly seek specialist care, or show that patients believe little long-lasting benefit is to be gained from physical/physiotherapy.

Participants had visited a range of other types of healthcare professional in the past year, including orthotists, podiatrists, and occupational therapists. Small proportions of participants had visited each professional, which suggests that the care requirements of CMT patients are varied.

Most participants had received rehabilitation therapies, medications, and aids/orthoses as part of their CMT care, and around half had undergone surgical procedures. The range of different CMT treatments received was wide.

Of note, high proportions of respondents had received pain medications for their CMT, and a relatively high number had also visited pain specialists. These results indicate that pain may have a substantial impact on people with CMT.

Another key observation is that many respondents reported use of aids/orthoses to help with walking, and had received surgeries to their feet or legs. This suggests that lower limb problems and mobility issues have a considerable impact on people with CMT.

Conclusions

Standards of care for CMT in the UK and US are broadly consistent with guidelines; however, there remains scope to improve access to suitable healthcare professionals.

Care is multifaceted, involving rehabilitation therapies, medications, orthoses and aids, as well as surgical procedures.

This ongoing study will provide further real-world insights into areas for the development of CMT care.

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