Care standards 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 standards of CMT management in UK and US real-world practice, including promptness of diagnosis and access to appropriate healthcare professionals.

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 the following aspects of their CMT care:

- Time to diagnosis
- Annual visit frequency to various healthcare professionals.

Outcomes were compared between countries and against UK and US CMT management guidelines.

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 care.

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)

Time to diagnosis

Time to CMT diagnosis is presented in Table 2.

Across all respondents, mean time from CMT symptom onset to diagnosis was 11.2 years. Mean time from first seeking medical care for CMT symptoms to diagnosis was 3.4 years.

Time to diagnosis was similar between countries. UK and US respondents reported mean times from symptom onset to diagnosis of 10.0 and 12.2 years, respectively, and from seeking care to diagnosis of 3.4 and 3.2 years, respectively.

Table 2: Time to diagnosis

Parameter	All respondents (n=361*)	UK (n=175)	US (n=179)
Mean age at symptom onset (years, SD)	19.3 (17.8)	20.3 (18.5)	18.0 (16.5)
Mean age at first seeking medical care (years, SD)	27.0 (17.9)	26.8 (18.7)	27.0 (16.8)
Mean age at diagnosis (years, SD)	30.4 (18.5)	30.3 (18.9)	30.2 (17.9)
Mean time from symptom onset to first seeking care (years)	7.7	6.6	9.0
Mean time from seeking care to diagnosis (years)	3.4	3.4	3.2
Mean time from symptom onset to diagnosis (years)	11.2	10.0	12.2

*Seven respondents did not report their country of residence. Abbreviations: SD, standard deviation

Annual healthcare professional visit frequency

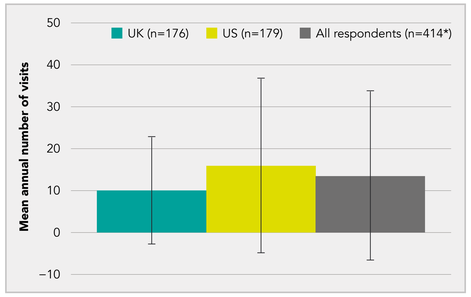
Annual visit frequency to CMT-related healthcare professionals is presented in Table 3 and Figures 1a and 1b.

Across all respondents (n=414), the mean annual number of visits to CMT-related healthcare professionals was 13.4 (standard deviation 23.4). Respondents visited a mean of 4.2 (standard deviation 1.1) different healthcare professionals.

There appeared to be some differences between countries. The mean annual number of visits to CMT-related healthcare professionals was numerically higher for US (15.8, standard deviation 29.2) than for UK respondents (9.9, standard deviation 9.4).

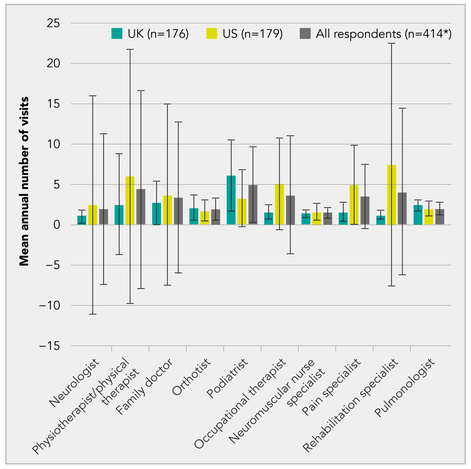
Across the majority of healthcare professionals, the mean number of annual visits was numerically higher for US than for UK respondents. Data distributions were generally wider for the US than for the UK.

Figure 1a: Annual healthcare professional visit frequency – total number of visits



*59 respondents did not report their country of residence. Error bars show standard deviation.

Figure 1b: Annual healthcare professional visit frequency – by professional



*59 respondents did not report their country of residence. Error bars show standard deviation.

Proportion of respondents visiting healthcare professionals annually

The proportions of respondents visiting each healthcare professional at least once a year are presented in Figure 2.

Core care team

Most respondents (71%) visited a neurologist at least once a year. The majority (74%) also visited their family doctor for problems with their CMT at least annually.

Physiotherapists/physical therapists were seen annually by 37% of respondents.

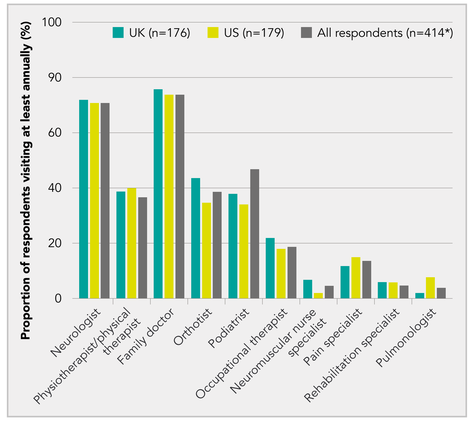
Wider care team

Orthotists and podiatrists were seen at least annually by relatively high proportions of respondents (39% and 47%, respectively).

However, only small proportions of patients (<20%) saw occupational therapists, pain specialists, rehabilitation specialists, and pulmonologists.

Similar proportions of respondents in each country visited each type of healthcare professional at least once a year.

Figure 2: Proportion of respondents visiting healthcare professionals annually



*59 respondents did not report their country of residence.

DISCUSSION

Diagnosis and care standards for CMT were generally aligned with guidelines[1-6].

However, respondents experienced average delays of several years from seeking care to receiving their CMT diagnosis. Respondents also reported a high mean time from symptom onset to seeking care. This may reflect poor awareness of CMT, leading sufferers to dismiss initial symptoms. Delays in seeking care and time to diagnosis were similar in the UK and US.

As recommended, the majority of respondents had at least yearly access to several members of a multidisciplinary care team. However, the type and number of healthcare professionals visited varied considerably:

- Most people visited a neurologist – the professional recommended to coordinate CMT care – at least once a year. However, a sizable minority did not.
- Physical therapists/physiotherapists, who are also core members of a CMT care team, were seen annually by less than half of respondents.
- The majority of people visited their family doctor at least once a year for problems with their CMT.
- The mean annual number of visits to family doctors was slightly higher than to neurologists.

These results may indicate that many people with CMT are well-managed by their family doctor, so do not regularly seek specialist care. However, they could also reflect limited access to specialists. Further investigation is warranted.

The proportions of respondents visiting each healthcare professional at least annually were similar in the UK and US. Some differences were apparent in the mean annual number of visits to certain professionals.

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

CONCLUSIONS

CMT care standards in the UK and US are broadly in alignment with guidelines; however, there may be scope to improve time to diagnosis and access to specialist care team members.

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

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Table 3: Healthcare professional visit frequency

Average annual visit frequency	Core care team			Wider care team							Total	Number of different professionals visited
	Neurologist	Physio/physical therapist*	Family doctor	Orthotist	Podiatrist	Occupational therapist	Neuromuscular nurse	Pain specialist	Rehabilitation specialist	Pulmonologist		
Mean (SD)	1.8 (9.4)	4.3 (12.3)	3.3 (9.4)	1.8 (1.4)	4.9 (4.7)	3.6 (7.4)	1.4 (0.7)	3.4 (4.0)	4.0 (10.4)	1.9 (0.8)	13.4 (23.4)	4.2 (1.1)
Median	1.0	0.0	1.0	1.0	4.0	1.0	1.0	1.0	1.0	2.0	7.0	4.0
Range	0.0–180.0	0.0–104.0	0.0–100.0	0.0–10.0	1.0–25.0	0.0–60.0	0.5–3.0	0.0–20.0	0.5–50.0	1.0–4.0	0.0–280.0	1.0–8.0

*Physiotherapists and physical therapists have very similar roles. Physiotherapist is the preferred term in the UK, while physical therapist is preferred in the US. Abbreviations: SD, standard deviation

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