

Digitally assessed real-world diversity in people with Charcot-Marie-Tooth disease in the UK and US

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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 from patients may therefore provide valuable insights.

OBJECTIVES

The objectives of this analysis were to examine the demographics of UK and US participants in a real-world observational study of CMT, and to investigate differences between those who had and had not participated in clinical trials.

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 interim analysis (data cut 30 May 2019, approximately seven months into the study) examined participants' responses to in-app surveys asking about the following demographic characteristics:

- Key demographics: country of residence, sex, age, body mass index
- CMT subtype
- Lifestyle: smoking status, exercise frequency, diet, alcohol intake
- Work/study status.

Differences between participants who had and had not taken part in clinical trials were explored.

Note: as most surveys in the app were optional and completed independently, different participants may have responded to each survey. Therefore, in this analysis, 'n' numbers vary by survey.

RESULTS

Demographics

Demographic characteristics for 666 survey respondents are presented in Table 1.

The proportions of respondents from the UK and US were similar.

Around two thirds of respondents were women. This was consistent across both countries, with 66.7% of UK respondents and 69.9% of US respondents being female.

The proportions of respondents aged <50 and ≥50 years were similar. This was consistent across both countries, with 53.0% of UK respondents and 49.4% of US respondents aged <50 years.

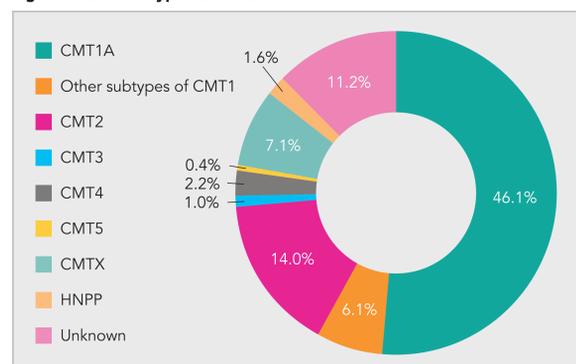
Table 1: Demographics

Parameter	Value
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)
Body mass index (mean, SD)	25.5 (11.4)

CMT subtype distribution

CMT subtype distribution for 492 survey respondents is presented in Figure 1. The most commonly reported subtype was CMT1A (46.1%), followed by CMT2 (14.0%), and Unknown (11.2%).

Figure 1: CMT subtype distribution



Lifestyle

Lifestyle characteristics for 404 respondents are presented in Table 2.

The majority of respondents (90.1%) were non-smokers.

Most respondents (84.2%) reported doing some exercise, with 61.4% exercising at least once a week. However, a notable minority of respondents reported that they were unable to exercise because of disability (13.4%).

The majority of respondents (60.9%) reported eating a varied diet, which included daily fruit and vegetables. A small minority (2.7%) reported eating a special medical diet. The most common medical diet was ketogenic (36.4% of those with a medical diet).

Most respondents reported low alcohol consumption, with 34.7% never drinking, and 46.5% drinking less than 1 unit per day.

Table 2: Lifestyle

Parameter	Respondents (n, %)
Smoking status	
Smoker	40 (9.9)
Non-smoker	364 (90.1)
Usual exercise frequency	
Daily	51 (12.6)
4–6 times per week	46 (11.4)
3 times per week	80 (19.8)
Once a week	71 (17.6)
Once a month	15 (3.7)
Seldom	77 (19.1)
Cannot exercise because of disability	54 (13.4)
Cannot exercise for other reasons	10 (2.5)
Usual diet	
No meat, fish or dairy produce	9 (2.2)
No meat	18 (4.5)
A mixture, including daily fruit and vegetables	246 (60.9)
A mixture, including regular chips and fries	49 (12.1)
Frozen meals 3–4 times per week	11 (2.7)
Mainly snacks	13 (3.2)
Medical diet	11 (2.7)
Usual alcohol intake	
Never drink alcohol	140 (34.7)
Less than 1 unit per day	188 (46.5)
1–2 units per day	61 (15.1)
3–6 units per day	10 (2.5)
7–9 units per day	4 (1.0)
More than 9 units per day	1 (0.2)

Work/study status

Work/study status for 412 respondents is presented in Table 3.

Around half of respondents (54.4%) were working for pay or studying. A considerable proportion (23.5%) were not working due to disability.

Table 3: Work/study status

Work/study status	Respondents (n, %)
Working (for pay)	205 (49.8)
Studying	19 (4.6)
Unemployed	23 (5.6)
Retired	68 (16.5)
Not working due to disability	97 (23.5)

Clinical trial participation

Twenty-three (5.9%) of 389 respondents reported having taken part in a clinical trial for CMT.

A demographic comparison of respondents who had and had not participated in clinical trials is presented in Table 4:

- The proportions of respondents from the UK and US differed between trial participants (UK 60.9% vs US 39.1%) and non-participants (UK 47.0% vs US 53.0%).
- The proportions of women and men were similar between trial participants (65.2% female vs 34.8% male) and non-participants (68.3% female vs 31.7% male).
- The mean age of trial participants was similar to that of non-participants (52.5 years vs 48.2 years, respectively).
- The proportion of people aged <50 years was slightly lower among trial participants (34.8% <50 vs 65.2% ≥50) than among non-participants (50.5% <50 vs 49.5% ≥50).
- The distribution of CMT subtypes was similar between trial participants and non-participants. CMT1A was the most common subtype in both groups (52.2% in trial participants vs 47.3% in non-participants), followed by CMT2 (26.1% in participants vs 14.2% in non-participants).

It should be noted that these results must be interpreted with caution given the small sample size of the trial participant group.

Table 4: Comparison of clinical trial participants and non-participants

Parameter	Trial participants (n=23)	Non-participants (n=366)
Country of residence (n,%)		
UK	14 (60.9)	172 (47.0)
US	9 (39.1)	194 (53.0)
Sex (n,%)		
Female	15 (65.2)	250 (68.3)
Male	8 (34.8)	116 (31.7)
Age (years)		
Mean	52.5	48.2
Standard deviation	14.7	13.9
Age <50 (n,%)	8 (34.8)	185 (50.5)
Age ≥50 (n,%)	15 (65.2)	181 (49.5)
CMT subtype (n,%)		
CMT1A	12 (52.2)	173 (47.3)
Other subtypes of CMT1	1 (0.0)	23 (6.3)
CMT2	6 (26.1)	52 (14.2)
CMT3	0 (0.0)	3 (0.8)
CMT4	0 (0.0)	9 (2.5)
CMT5	0 (0.0)	1 (0.3)
CMTX	1 (4.3)	29 (7.9)
HNPP	0 (0.0)	6 (1.6)
Unknown	3 (13.0)	38 (10.4)
Not reported	1 (4.3)	32 (8.7)

DISCUSSION

Demographic diversity was high in this digital real-world study of CMT. Participants were relatively evenly distributed across countries and age groups.

The proportion of women enrolled was high. As it is generally accepted that the prevalence of CMT is not sex-dependent, apart from a high occurrence of CMTX in men^{1,2}, this finding may reflect sex-related differences in the likelihood of enrollment, e.g. a higher willingness to participate among women.

The distribution of CMT subtypes was similar to that observed elsewhere, with CMT1A, other subtypes of CMT1, CMT2, and CMTX being the most common subtypes among participants who knew this information^{3,4}.

The majority of participants reported having a reasonably healthy lifestyle – not smoking, taking regular exercise, eating a varied diet, and consuming low levels of alcohol.

Only around half of participants were working or studying. The remainder were retired, unemployed, or not working due to disability. Investigation of the potential effect of CMT on work/study status is ongoing as part of this study.

A small proportion of participants had taken part in a clinical trial for CMT. Demographic characteristics were generally similar between those who had participated in trials and those who had not.

The objective of this ongoing observational study is to understand the impact of CMT in the real-world, and in the broad range of patients encountered in clinical practice, rather than in the heavily preselected patients who are generally eligible for clinical trial participation. The demographic diversity observed in this interim analysis is therefore promising.

CONCLUSIONS

Demographic diversity was high among UK and US participants in this observational study of CMT.

Demographic characteristics were similar between those who had and had not participated in clinical trials.

An important finding was the high proportion of participants who were not working or studying. Investigation of the potential effect of CMT on work/study status and productivity loss is ongoing as part of this study.

This ongoing study will provide further real-world insights to increase understanding of CMT.

REFERENCES

- 1 Charcot-Marie-Tooth UK. Health professionals. Epidemiology. 2019. Available at: <https://www.cmt.org.uk/health-professionals/>. Accessed: Jun 2019
- 2 Medscape. What are the sexual predilections of Charcot-Marie-Tooth (CMT) disease? 2018. Available at: <https://www.medscape.com/answers/1173104-157205/what-are-the-sexual-predilections-of-charcot-marie-tooth-cmt-disease>. Accessed: Jun 2019
- 3 Fridman V et al. J Neurology Neurosurg Psychiatry 2015;86:873–8
- 4 Pareyson D et al. Curr Opin Neurol 2017;30(5):471–80

