Demographics and treatment patterns for Charcot-Marie-Tooth disease in the EU and US: interim results from an international digital real-world evidence 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 choices2. There is currently no curative treatment, meaning that patients are reliant on supportive care.

CMT is rare, and – until recently – there has 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 objective of this analysis was to examine patient-reported demographics and treatment patterns for CMT in the EU and US real-world setting.

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 demographic characteristics, CMT management, and quality of life.

This interim analysis (data cut 27 August 2019, approximately ten months after recruitment began) examined demographics and CMT treatment patterns reported by participants from Germany, Italy, Spain, the UK, and the US.

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 may vary by survey.

Results

Demographics

Key demographics for the 953 respondents to this survey are presented in Table 1.

As initiation of recruitment was tiered by country, at the time of analysis, the distribution of study participants across countries was uneven, with most respondents living in the UK and US.

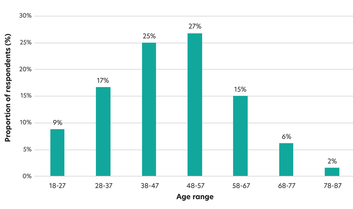
Around two thirds of respondents were women.

Respondent age distribution is presented in Figure 1. Respondent age was well-distributed.

Table 1: Key demographics

Parameter	Value
Country of residence (n, %)	
Germany (recruitment began January 2019)	74 (7.8)
Italy (recruitment began January 2019)	141 (14.8)
Spain (recruitment began April 2019)	71 (7.5)
UK (recruitment began November 2018)	330 (34.6)
US (recruitment began October 2018)	337 (35.4)
Sex (n,%)	
Male	630 (66.1)
Female	322 (33.8)
Age, years	
Median (range)	47 (18-83)

Figure 1: Age distribution



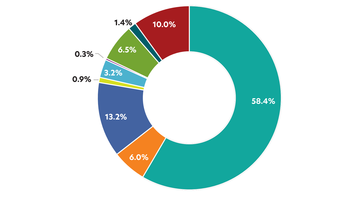
CMT characteristics

CMT subtype distribution for 770 respondents is presented in Figure 2.

The most common subtype among respondents who knew this information was CMT1, followed by CMT2 and CMTX.

In addition to CMT, 21.2% of respondents reported having being diagnosed with at least one other medical condition.

Figure 2: CMT subtype



Treatment patterns

Figures 3 to 6 present the proportions of respondents (n=734) who reported having ever received medications, rehabilitation therapies, aids/orthoses, or surgical procedures as part of their CMT care, respectively. Within each treatment category, respondents were able to select multiple treatments.

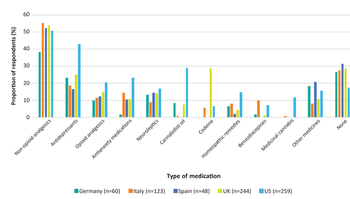
The majority of respondents (69-83%) in all countries had received medication for their CMT symptoms.

The proportion of respondents having received medication was highest in the US, closely followed by Germany, Italy, and the UK.

The most commonly used medications across the respondent population as a whole were non-opioids analgesics, antidepressants, and opioid analgesics.

Reported use of antidepressants, cannabidiol oil, and medicinal cannabis was higher in the US than in Europe.

Figure 3: Proportion of respondents who received different types of medications

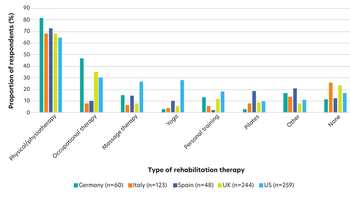


The majority of respondents in all countries (74-88%) had received rehabilitation therapy for their CMT.

The proportion of respondents who had received physical therapy was highest in Germany, followed by Spain, and the UK.

Across the respondent population as a whole, physical/physiotherapy, occupational therapy, and massage therapy were the most commonly reported therapies.

Figure 4: Proportion of respondents who received different types of rehabilitation therapy

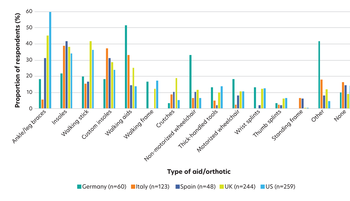


Most respondents (84-90%) in all countries had used aids or orthoses for their CMT.

The countries with the highest proportions of users were Germany and the UK, with Italy, Spain and the US having slightly lower proportions.

The most commonly used aids or orthoses across the respondent population as a whole were ankle/leg braces, insoles, and walking sticks.

Figure 5: Proportion of respondents who received different types of aids/orthoses

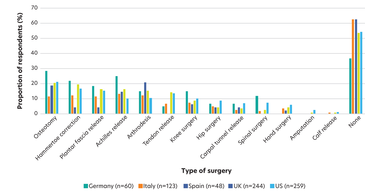


The proportion of respondents who had undergone surgery for their CMT was notably lower than the proportions who had received other treatments.

Surgery was most common in Germany, with almost two-thirds of respondents having underdone a procedure for their CMT, and lowest in Italy and Spain, where only around a third of respondents had received surgery.

Across the respondent population as a whole, the most common procedures were osteotomy, hammertoe correction, and plantar fascia release.

Figure 6: Proportion of respondents who received different types of surgery



Discussion

The demographic diversity observed in this real-world study of CMT patients was high, suggesting that data collected in the study are likely to be representative of, and generalizable to, the wider CMT patient population.

Participants were well-distributed by age. Geographic distribution was skewed, but this reflected the ongoing state of recruitment.

The proportion of women enrolled was high. As it is generally accepted that the prevalence of CMT is not sex-dependent, apart from a higher occurrence of CMTX in men[3,4], this finding may reflect sex-related differences in the likelihood of enrollment, i.e., women may be more willing to participate in this kind of research than men.

The distribution of CMT subtypes was similar to that observed elsewhere[5,4], with CMT1A, CMT2, and CMTX being the most common subtypes among participants who knew this information.

Across all countries, most participants had received medications, rehabilitation therapies, and aids/orthoses for their CMT. Use of surgery was less common in all countries. Overall, one fifth of participants reported receiving no rehabilitation therapies; suggesting gaps in CMT management. Although broad approaches to CMT care appeared consistent among countries, there was some variation in the specific treatments used.

High proportions of respondents had received some form of pain medication for their CMT, suggesting that pain may have a substantial impact on people with CMT. In addition, many respondents used aids/orthoses that provided assistance with walking, and had received surgeries to their feet or legs, which indicates that lower limb problems and mobility issues may have a considerable impact on people with CMT.

The proportions of participants who had received each type of treatment were consistently high in Germany. This aligns with high reported frequencies of healthcare visits in Germany observed in another analysis from this study[7], and may reflect an intensive approach to CMT management, or a high proportion of people with severe CMT.

Conclusions

The management of CMT is multifaceted, involving the use of medications, rehabilitation therapies, aids/orthoses, and surgical procedures.

Although broad approaches to treatment appear to be consistent across the US and EU, the choice of individual treatments does seem to vary by country.

This ongoing study will provide further real-world insights into CMT treatment to enable gaps in care to be identified and addressed.

References

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