

Health-Related Quality of Life of Patients with Charcot–Marie–Tooth Disease: A Systematic Review

Authors: Sukannya Mahapatra, Nidhun Kandoth, Vyshnavi Telukuntla, Amit Ahuja, Inderpreet Singh Khurana

Affiliations: Lumarity, Gurugram, HR, India

INTRODUCTION

- Charcot–Marie–Tooth disease (CMT) is one of the most prevalent hereditary neurological illnesses, affecting roughly 1 in 2,500 persons worldwide¹
- It includes a range of genetically and clinically diverse disorders that are characterized by a persistent, increasing peripheral nerve impairment²
- The disease manifests with varying degrees of severity and can significantly impact the quality of life (QoL) of affected individuals³

OBJECTIVES

We aimed to systematically examine the extent to which CMT affects QoL, utilizing widely recognized QoL measurement tools.

METHODS

- A systematic literature search was conducted in the Embase® and Medline® databases in adherence to the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines, to identify articles that reported data on health-related quality of life (HRQL) and utility related to CMT
- The search strategy employed relevant keywords to identify English-language studies focusing on the HRQL and utility of patients with CMT
- Inclusion criteria were pre-defined, with a specific emphasis on studies reporting HRQL and utility data for adult patients with CMT (Table 1)
- Two independent reviewers initially screened the titles and abstracts of all records identified through the electronic search. Subsequently, potentially relevant full-text articles were assessed by both the reviewers
- In case of disagreement or uncertainty regarding inclusion, a third reviewer was consulted to reach a consensus

Table 1. Inclusion criteria

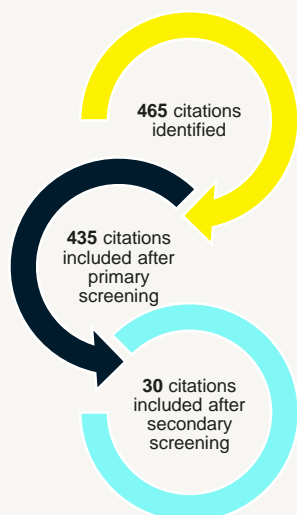
Population	Patients diagnosed with CMT
Population	Patients diagnosed with CMT
Outcomes	<ul style="list-style-type: none"> Results from any HRQL instruments Patient and caregiver HRQL Utility values Patient-reported outcomes Patient preference
Language	English
Study design	<ul style="list-style-type: none"> Observational studies RCTs (reporting relevant outcomes) Case-control studies Cross-sectional studies Utility studies (including studies where utility weights were mapped from other instruments, e.g. disease-specific patient reported outcome measures) Utility estimates reported in economic evaluations
Time frame	Inception–present (2024)
Language	English
Publication type	Full-text articles, conference abstracts
Intervention and comparator	No restriction
Country	No restriction

Keys: CMT, Charcot–Marie–Tooth disease; HRQL, health related quality of life; RCT, randomized controlled trial.
Note: No restriction was applied for the age of patients with CMT.

RESULTS

- The systematic review identified a total of 465 studies, of which 30 met the inclusion criteria for our qualitative analysis (Figure 1)

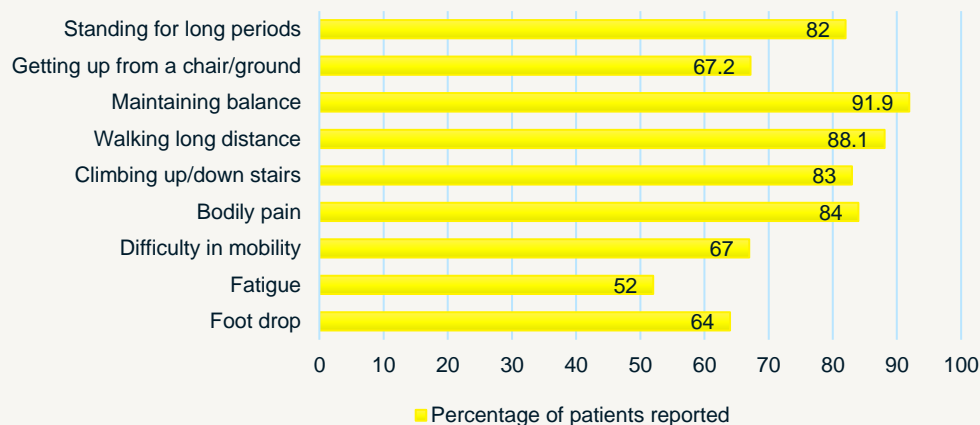
Figure 1. Study flow diagram



- Most studies were conducted in European countries (n = 8), the US (n = 8), globally (n = 8), Australia (n = 3), Brazil (n = 2), and Serbia (n = 1)
- The EQ-5D-5L and 36-Item Short Form Survey (SF-36) were the most frequently administered QoL scales

- In patients with CMT, the primary symptoms contributing to QoL deterioration were maintaining balance (91.9%), walking long distance (88.1%), bodily pain (84%), difficulty in mobility (67%), fatigue (52%), and foot drop (64%) (Figure 2)^{4,5}
- Additionally, 50% of patients described their pain as shooting, stabbing, burning or severe, with the main pain sites being the feet, legs, knees and lower back⁶

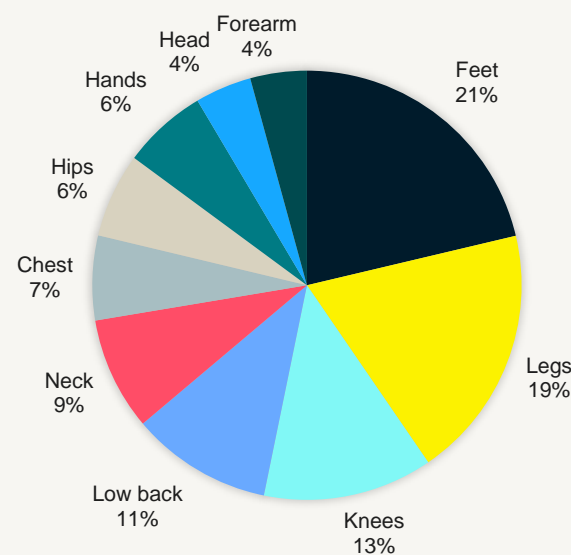
Figure 2. Major contributing symptoms to QoL deterioration



Key: QoL, quality of life.
Source: Nussbaum and Moore. *Muscle Nerve*. 2017; 56(3), Moore et al. *J Peripher Nerv Syst*. 2017; 342-3.

- The most common pain locations for patients with CMT were reported to be the feet, legs and knees (Figure 3)⁷
- A study based in the US found significant correlations between the SF-36 physical functioning domain and pain intensity on the visual analogue scale (VAS) ($p < 0.01$), as well as between limitations due to physical health and the VAS ($p < 0.004$)⁸
- A study conducted in France reported mean (standard deviation [SD]) scores for physical and mental SF-36 domains of 36.4 (10.0) and 48.4 (11.5), respectively⁹
- Age and disease severity were identified as the primary factors exacerbating QoL in patients with CMT^{10,3}
- Aging exacerbates muscle loss and weakness, leading to reduced mobility and increased difficulty in performing daily activities. Progressive sensory loss with age can cause balance issues and fine motor skill difficulties, increasing injury risk^{10,3}

Figure 3. Frequency of pain experienced by patients with CMT in different locations



Key: CMT, Charcot–Marie–Tooth Disease.
Source: Azevedo et al. *Arq Neuropsiquiatr*. 2021; 79:415-9.

CONCLUSIONS

- Pain is a predominant and critical symptom in CMT
- Our findings emphasize the need for targeted interventions to manage pain and mobility issues, as well as comprehensive care strategies that address the multifaceted nature of QoL deterioration in individuals with CMT

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