

## Deliverable D.4.1.

# Towards Rehabilitation Guidance for the Management of Adults Living with Charcot-Marie-Tooth Disease

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As a rare inherited neuromuscular disease CMT is scarcely known among Health Professionals. Medical prescriptions for physiotherapy and other rehabilitation interventions are often inappropriate and not appropriate in some healthcare contexts where Physiotherapists and other Allied Health Professionals work autonomously. Many rehabilitation professionals lack knowledge of the specific needs of CMT patients. Information for people living with CMT is sparse and it is difficult to find good quality guidance. Guidelines would not only help to instruct Health Professionals who deliver rehabilitation but also be useful references for people living with CMT who are trying to self-manage. This would not only benefit the people living with CMT to manage their disease but also alleviate the financial burden on the healthcare system.

Participants of the 2<sup>nd</sup> European CMT Specialists Conference October 2025 in Antwerp learned that guidelines for managing CMT do already exist for children in English, Spanish and Portuguese (de Paula et al., 2024), and it was agreed that they should be translated into other languages and so be made available to a broader group of patients. Meanwhile, translations into Italian and into German have been prepared in cooperation with ECMTF and are about to be published.

However, an appropriate equivalent of these guidelines for adults has yet to be elaborated (Burns et al., 2026). Thus, the idea was born that a group of researchers specialized in the symptomatic treatment and rehabilitation of people living with CMT initiate the development of rehabilitation guidance for the management of adults living with the Charcot-Marie-Tooth Disease. The following thoughts outline the situation and some first steps envisaged to turn the idea into reality:

## 1. Background

CMT is a hereditary neuromuscular disease, with more than 100 possible genetic mutations found to date, causing peripheral nerve degeneration (Laurá et al., 2019; Reilly and Rossor, 2020). Most commonly, CMT presents with varying degrees of slowly progressive, length-dependent neuropathy, manifesting in distal weakness and sensory loss. These symptoms present significant challenges for people living with CMT that can include problems with walking, balance, hand function, pain, foot deformity, fatigue, nutrition, respiratory, voice and psychological burden.

The World Health Organisation define rehabilitation as a set of interventions designed to optimize functioning and reduce disability in individuals with health conditions, in interactions with their environment (WHO World Health Organisation, 2024). Four types of rehabilitation have been described that could apply to people living with CMT across the lifespan:

- **Preventative:** maintaining function for as long as possible
- **Restorative:** after surgery, illness or acute events to maximise function
- **Supportive:** increases abilities through methods such as providing self-help devices, compensatory strategies, assistive equipment or environmental modifications
- **Palliative:** maximise functional independence and support comfort, dignity and quality of life

(Herbert Dietz, 1969)

To meet these goals, people living with CMT benefit from access to multi-disciplinary rehabilitation from a range of Health Professionals to support them to live well, and this should be available across different context of how care is delivered in different countries and healthcare systems internationally.

Evidence-based guidelines to support rehabilitation are required and where there are evidence gaps, guidance can be developed with consensus from experts in the management of CMT, and in partnership with people living with CMT as experts from experience in all stages of guideline development.

In other conditions, even where rehabilitation recommendations exist, translation into routine care is variable as clinicians and services face barriers such as limited time, workforce capacity, training, and access to MDT inputs (e.g., orthotics/OT/psychology), meaning guidance may not reach people living with CMT consistently.

Models of CMT care differ widely across countries and regions (specialist vs generalist pathways, referral systems, funding/commissioning, availability of assistive technology), so guidance must be designed with a clear distinction between *core components* and *adaptable elements* to support equitable uptake.

Rehabilitation guidance is inherently a complex intervention involving multiple professions and coordinated care processes. Implementation therefore requires understanding how roles, workflows, and interprofessional communication influence uptake and sustainability in real-world settings.

To support real-world roll-out, guidance should be accompanied by practical implementation supports (e.g., training materials, templates/checklists, minimal audit indicators) co-developed with stakeholders, enabling services to monitor adoption and refine delivery over time.

## 2. Guidelines to date:

Paediatric CMT clinical practice guidelines have recently been published by an international consortium by combining systematic reviews, a modified Delphi and application of the Grading of Recommendations, Assessment, Development and Evaluation (GRADE) approach. However, the underpinning evidence base in children is limited, with only a small number of studies meeting inclusion criteria across the review questions, meaning that most recommendations are consensus-based rather than evidence-based.

The guideline provides three evidence-based recommendations, including support for progressive resistance exercise targeting ankle dorsiflexors, and conditional recommendations against high-dose ascorbic acid and lower-limb botulinum toxin for flexible cavus in CMT1A, alongside wider consensus recommendations spanning common rehabilitation-relevant issues such as weakness, mobility and balance, sensory symptoms and cramps, upper limb function, joint range, orthoses/footwear, deformity management and respiratory monitoring (Yiu et al., 2022).

In 2018, Physical Therapy and Occupational Therapy guidelines were developed from a consensus event, funded by the CMTA, harnessing expert opinion from physiotherapists and Occupational Therapists from the USA, UK and Italy (Charcot Marie Tooth Association., 2018). The event was informed by survey of people living with CMT in the USA, but there was no formal evidence synthesis.

Expert opinion guidance on rehabilitation options for people living with peripheral neuropathy included research on CMT, but this was an educational article (Ramdharry et al., 2020).

Not all current guidance has involved of people living with CMT in the development process, and there is no consideration of differing healthcare contexts for implementation of guidelines globally.

There needs to be an updated guide for management and rehabilitation interventions for adults living with CMT, conducted through rigorous methods of literature review, expert consensus and consideration of wide-reaching implementation internationally. Internationally renowned colleagues involved in the research of rehabilitation interventions for people living with CMT will be invited as co-investigators for this proposed work.

## 3. Methods

A three-stage approach to developing guidelines is envisaged to develop prototype guidance from evidence synthesis, patient engagement and expert opinion through consensus methods. This program of work will culminate in exploration of implementation factors for the guidelines into global practice, through seeking opinion from patients and colleagues from a range of countries and differing healthcare systems in high and low-to-medium income countries.

## 4. Deliverables & Dissemination

The work from the three stages will be published in peer reviewed journals and presented at relevant conferences. The research team will also engage with patient organisations and professional networks internationally to present and raise awareness of the guidelines.

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