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Outline for a digital care strategy for CMT across Europe and beyond

Helena F. Pernice¹, Maike Dohrn², Rafael Sivera Mascaró³, Maurizio D'Antonio⁴, Tim Vangansewinkel⁵, Filippo Genovese⁶

1. *Department of Neurology, Charité University Medicine, Berlin, Germany*
2. *Department of Neurology & Neuromuscular Center, Medical Faculty of the RWTH Aachen University, Pauwelsstr. 30, 52074 Aachen, Germany*
3. *Unidad de enfermedades neuromusculares y ataxias, Hospital Universitari i Politècnic La Fe, Valencia, Spain*
4. *IRCCS Ospedale San Raffaele, Milano, Italy*
5. *Department of Neuroscience, BIOMED Biomedical Research Institute, Faculty of Medicine and Life Sciences, Hasselt University, Hasselt, Belgium*
6. *European CMT Research Federation (ECMTF)*

T6.2 - Telemedicine in iNMD: Digital care and other aspects of telemedicine to provide for direct communication between patients and specialists - challenges and opportunities.

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ECRA's perspective on digital care: where are we now?

During the Antwerp conference, we discussed challenges in the current care landscape for CMT, which mainly suffers from a lack of specialized and accessible care and lack of awareness in primary clinical contacts of patients such as general practitioners or orthopedic specialists. Since CMT is a rare disease, patients often spend years to decades without a clear diagnosis, in spite of an odyssey of visiting different specialists and partially invasive diagnostic procedures. Even when the diagnosis is made thanks to the wider availability of genetic testing, little information or advice is given to patients: no cure exists, patients are often left alone with their destinies, leading to important psychological impact and a high private and public socioeconomic burden. Those CMT specialists available are often difficult to reach due to geographic distances, in particular for people with movement disabilities like CMT, and due to long waiting times (often several months), just to receive simple answers of how to behave or what to expect. Furthermore, understanding the disease and performing clinical studies for new treatments unconditionally depends on the inclusion of large patient numbers for researchers and pharmaceutical companies to plan promising trials. As patients are dispersed and often not connected to specialist centers, reaching these numbers and following up disease courses to better understand influencing factors is often near impossible in the rare disease landscape. Consequently, close international collaboration and innovative solutions are necessary to bridge these gaps in CMT care and research.

Digital care, such as tools for streamlined information, remote interaction with specialists or disease trackers, as well as digital networks for interdisciplinary care, can be the path towards a solution of these challenges: it would facilitate access to specialists for patients and local physicians; continued cost-effective consultation across distances and borders; systematic collection of health data and natural history as a basis of advanced, AI-supported research;

international cooperation in data-management and analysis. For rare disease care like CMT, in particular, digitalization of care remedies the scarce availability of reliable specialist knowledge and therapy. Past exploration of a smartphone application (CMT&Me) to inform patients and collect data remotely showed a promising example of such a tool, but was based only on one pharmaceutical company, which caused discontinuation and lack of data availability (1). An important aspect of future digital tools is to ensure sustainability beyond single funders and data availability to the whole research community.

The aim of this project is to better understand the problems that the CMT community – patients, researchers, healthcare professionals, and industry – face and to develop solutions within the digital field that provide real and sustainable value to all stakeholders. Digitalization of care is already being implemented in several member states and health systems EU-wide and beyond. However, few healthcare systems have reached a state of using digital tools in a daily and systematic fashion. National solutions do not seem sufficient. Common action is needed, striving towards a system of digital care across Europe for the benefit of patients, and cost-effectiveness of our health systems at large.

Specification of framework:

ECRA members are committed to operating on a global rather than European scale only. Given that ECRA is a European association, our aim is to overcome rather than create barriers. For the abovementioned reasons (rareness and diversity of CMT), a network structure will be most versatile and promising if it is open to foster international collaboration for maximum outcome.

Challenges for developing solutions in CMT care

Coming together as ECRA members from different countries, challenges in developing joint digital solutions have been identified.

1. **Differences between countries.** Due to the large diversity of health systems within European countries, also care for CMT patients differs greatly. During the conference, we presented key challenges in care within the patient journey in Germany. This evoked an important discussion revealing that in some countries, challenges are already being faced, primarily due to strong patient advocacy (Italy, France), while in other countries basic needs such as electronic health records are still lacking.
2. **Regulatory barriers.** Despite existing efforts to homogenize care access across Europe, many countries still impose strict and individual regulatory requirements for any digital tools used in healthcare. Especially tools with a potential impact on medical decisions are strictly regulated and face long approval processes that differ between countries. This includes data protection regulation and security: telemedical systems that are used in one country, may not be available in others. During our discussion in the Antwerp conference, we identified that key drivers for regulatory and political changes necessary to implement new care structures are the illumination and understanding of the socioeconomic impact of the disease. While specialists are well aware of the high healthcare and indirect costs associated with the disease and some studies on quality of life and single-country economic aspects have been performed (1-6), there is a profound lack of up-to-date, international, disease-specific analyses of the socioeconomic impact on health systems at large.
3. **Access and language barriers.** We further identified that in some countries and ethnicities, access to digital tools is restricted by availability or believes. For example, in

Romania, people may not be acquainted with internet-based structures or simply prefer analog and face-to-face care. If whole care structures are digitalized, such societies may be left out. Furthermore, when discussing solutions that already exist (elaborate information material from CMT-USA and comic illustration of the disease from CMT-France) we found that they cannot easily be disseminated due to language barriers and would need individual translations, which have to be carefully revised by specialists to maintain content quality.

To overcome these challenges, ECRA has developed the following work steps:

- 1) Better understand the patient journey in CMT across countries
- 2) Better understand economic impact of CMT across countries
- 3) Develop joint solutions for overcoming identified needs using existing and new digital tools for international use

For **workstep 1**, we heard presentations on digital care strategies in different countries during the Antwerp conference. We explored the challenges and opportunities of digital solutions being developed in Germany as an example. We here reached the conclusion, that there is an important knowledge gap of patient needs across countries. In the proposition and discussion of a future ECRA Joint Project, we outlined the development of a questionnaire for patients across different countries to better map needs and challenges in care. Here, we aim to closely collaborate with the ERN Euro-NMD, who are currently formulating a patient-journey poster for CMT by collecting viewpoints of different stake holders (Arabella Acalinei). Furthermore, we came together as a task force (Filippo Genovese, Alexandre Hoyau, Nadège Costa, Arianna Fornari, Davide Pareyson) to share currently planned and ongoing projects for national patient journey surveys to provide these for other countries. A joint project proposal for homogenized surveys across countries is envisioned in this first workstep.

During the Antwerp conference, we identified that, indeed, economic aspects are the largest driver for any efforts in improving healthcare structures. For this study we intend to closely collaborate with the CureCMT Joint Project led by Kleopas Kleopa, which includes an analysis of the economic impact of the disease. It will be important to bring together experienced and junior researchers to review and compare of available data on healthcare utilization, direct and indirect costs, access to care, employment, education, and caregiver burden, incorporating both published evidence and expert country-level insights. **Workstep 2** intends a collaborative analysis to be conducted, cross-country synthesis to identify disparities, data gaps, and priority areas for future research and policy action.

Workstep 3, planned for Q4 2026 and Q1 2027, incorporates the formation of an international task force to collect existing digital solutions and identify missing features from worksteps 1 and 2. Then, a Joint Project application for funding for the development of a prototype for a digital tool or app is planned. Next steps will include field testing and usability of the tool with the different stake holder groups (patients, healthcare professionals, industry).

Conclusion

ECRA's **strategy for digital care**, thus, comprises three steps as outlined above: a detailed understanding of all stakeholder needs in the CMT community, an explicit mapping of the current economic impact and pointless wasted expenses, due to malfunctioning care strategies across countries, and an outline of concrete tools and solutions available or needed to be developed in a joint r&d project based on the knowledge acquired during our planned worksteps.

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