

## Deliverable D.6.2.

# Cooperative Research & Development for a Cure of CMT - “patients as partners”

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### 1. The concept: Patients as Partners

A new concept has already governed the 1st European CMT Specialists Conference 2023 in Paris: “patients as partners”. For a first time a patient organization - namely the European CMT Federation (ECMTF) - has taken the initiative to bring together scientists, clinicians and other health professionals to a joint conference, determined to take CMT-research to a new, more effective, level and so create conditions for rapidly finding a cure to this threatening disease. Professional specialists and those living with the disease could not only share their respective expertise - medical, biological, chemical etc. science on the one hand, and living with the many variants of CMT, on the other - and learn from each other but also discuss strategies for boosting research in this field, on equal terms.

- The **first outcome** of this conference was the awareness that this innovative and very successful joint venture should be further developed two years later, with the 2<sup>nd</sup> European CMT Specialists Conference. This conference took place in October 23-25, 2025, in Antwerp.
- The **second outcome** was a decision to establish a European CMT Research Association (ECRA) as a joint initiative of the patients (ECMTF) and the professional CMT specialists. Thanks to the intense and committed cooperation of the ECRA taskforce, consisting of leading scientists and patient representatives, this association was established in fall 2024. Both organizations together undertook to jointly organize the 2<sup>nd</sup> European CMT Specialists Conference, and this in close partnership with the University of Antwerp that also hosted the conference.
- As a **third outcome**, it was decided in Paris that a patient-owned CMT health data management system shall be established in response to the particular need of joining all efforts and capacities to provide easy and, if possible, free access to all researchers to the patients data that are indispensable for effective research, especially in the age of artificial intelligence.

ECRA and the Antwerp Conference took the concept up and developed it further as follows:

## 2. Rethinking a Special Relationship

A **fundamental rethink** is necessary not only among scientists and physicians, but also among patients. The Antwerp Conference has shown that it is possible, all participants have experienced that the new concept “patients as partners” is beneficial to all, including the industry, whose task is to turn the findings of the researchers into practical application: a cure for CMT and other inherited neuromuscular diseases. The cooperation to be promoted, indeed, includes the industries, they are part of the game, from the scratch, their early engagement (expertise and funding) in joint research projects promises straight, and targeted research.

The **first rethink is for the patients**: As discussed in Paris and successfully practiced already in Antwerp, patients are not the subject of scientific and medical efforts only, not purely passive consumers of services, dependent of the masters’ help; only if they engage, collaborate actively in the processes of research (e.g. with precise accounts of their natural history), of clinical and of other therapeutic care (e.g. regular exercises, regular consultation with the doctor and monitoring the progress of the disease), but also of the development of therapies and drugs (e.g. clinical trial readiness), all this as partners of their professional counterpart - only with this personal engagement they can expect that substantial progress be made in finding their so urgently hoped-for cure. Patients need to be mobilized in this sense.

The **second rethink is for the professionals**: scientists, clinicians, therapists, and even the caregivers learn and accept that their efforts can become significantly more effective if they treat their patients on equal terms, as partners in the joint venture of healing. It was rightly asked at the Antwerp Conference if it is (still) appropriate at all to use the term “patient”. Etymologically the word seems to imply suffering, passively enduring pain or discomfort on the one side, kindness and understanding of the caregiver on the other side. Even where, today, the term is substituted by client, consumer or person who receives care, the idea of active partnership is not present. Talking about a person affected by this or that disease might be more appropriate and leaves room for the idea of partnership. Scientists and health professionals viewing the people affected with the disease as their partners, with the appropriate respect, might release great forces, creativity and readiness of cooperation as is needed for effective research and treatment.

## 3. Preparing the Discussion

To prepare the Antwerp Conference the first webinar of the “**Pre-Antwerp Conference Webinar Series**” launched in May 2025 (<https://www.uantwerpen.be/en/conferences/2nd-european-cmt-specialist-conference/webinars/webinar-1/>) took the subject up with a conversation on

- Patients in Action: Discover what CMT patient organizations offer to you as a patient.
- Empowering Patients: Learn what patients can do for their organizations, and
- Patients as Partners: Understand the concept of “patients as partners” and its validity.

Discussing the concept of “patients as partners” could become too theoretical if it is not based upon facts. To provide participants of the Conference at least with a first idea of what patients are ready to do about their disease and the relationship with the profession,

a survey was conducted in summer 2025 among patient organizations, titled “**Patient Readiness for Partnership in Research on inherited NeuroMuscular Diseases (iNMD)**”. The report of the survey is published at the Conference website (<https://www.uantwerpen.be/en/conferences/2nd-european-cmt-specialist-conference/materials/other/>).

The results are positive and encouraging: (a) Patients do wish to have more information on the disease and of the progress of research in the field; (b) patients are willing to closely cooperate with scientists and medical professionals in order to halt the progress of their disease; and (c) patient organizations play an important role as an interface between patients and the medical profession.

#### **4. A Practical Example: Patients as Partners in Research (CMTA)**

Katherine Forsey, the CSO of the powerful American patients organization CMTA, member of both, ECMTF and ECRA, gave an extremely insightful and inspiring report at the Antwerp Conference on “**Patients as Partners in Research: The CMTA Experience**”, available at: <https://www.youtube.com/watch?v=bncq-23YDTg>. In order to allow patients to be involved in the research process, on the one side, and allow researchers to find patients where appropriate and to connect with them to push forward their research work, CMTA launched in 2018 its “patients as partners in research” platform. With over 8700 patient profiles from all over the world in October 2025, and the number is rapidly growing, making it the largest patient reported outcome measures dataset worldwide. It allows to find CMT patients for studies and for clinical trials, to easily organize surveys on the different types of CMT and to engage in education, and information of the patients on new findings in research and new opportunities to participate in research projects.

#### **5. Conclusion: Strategy towards Partnership**

The new approach of partnership between patients and health professionals at large, including scientists, clinicians, therapists and care givers requires openness on both sides. Mobilizing patients to become active partners would be one task, encouraging the profession to listen and accept patients with the contribution they can give to improve the effectiveness of their treatment and the research efforts in the field is another.

Discussing and even practicing the concept at conferences like the Antwerp Conference will not be sufficient to spreading the idea among the public, as needed, and putting it into practice. To organize great conferences on patients as partners in clinical research, as planned for Boston in March 2026 (see: <https://patientsaspartnersconference.com>) may be a useful tool for exchanging experiences and finding new ways to mobilizing patients, but if the outcome justifies the high investment and registration fees even for patient advocats remains to be seen.

So far, facing the challenges of iNMD’s and CMT in particular, three steps should be considered to make practical progress towards real partnership in research for the benefit of all stakeholders involved:

- a. Only a little part of CMT patients are aware of the cause and origine of their slowly progressing disability, many even tend to avoid talking about and taking measures to do something about it - until it becomes impossible to ignore and hide. The result is that effective (symptomatic) therapies already available at

an early stage are missed and progress of the disease is not delayed as it could be. **Awareness campaigns** regularly conducted by ECMTF help overcome the fear and obstacles against accepting the diseases and taking action as required to mitigate the consequences. Awareness campaigns should include the partnership idea and give advice how to actively engage in a joint effort with the health profession to timely do what is possible.

- b. Such campaigns would also raise awareness among health professionals, not only of the very existence of these rare diseases but also of the opportunities that the partnership approach offers for making therapeutic and research efforts more effective. **Practical examples of successful cooperation** as given in the presentation referred to supra 4. help making the benefits of the concept more plausible. A **survey** on researchers', but also of patients' positive experience should be performed, and the results should broadly be published. **A movie** to be produced of a telling case of living with CMT would substantially add to awareness of the diseases but should include the partnership concept as a guideline for how to improve therapies and accelerate the research process towards finding a cure on CMT.
- c. The experience with the 1<sup>st</sup> and the 2<sup>nd</sup> European CMT Specialists Conferences indicates that patient-initiated scientific conferences are well received by the professionals: an extremely productive and open-minded cooperation led to a number of new joint research projects and innovative initiatives. Yet, to organize such gatherings is burdensome and brings about so far unknown challenges. Patients and members of their organizations often do not dispose of a secretariat to assist the administrative work of preparing a conference and fundraising, nor do they have the funding to cover the traveling and accommodation costs to participate in person. Other sources, therefore, must be found for **financing such events** if they are supposed to be on a partnership basis. Private sponsorship can only serve in part. The solution found was to apply for support under EU programs such as EU4Health, but the time and effort involved for the grant application and reporting requirements after the conference proved to be completely disproportionate in relation to the final yield. A new kind of funding instruments, thus, needs to be established, particularly to support promising initiatives like boosting research in the field of rare diseases, based upon the "patients as partners" concept.

## Publications:

### 1. Conceptual Frameworks: "Patients as Partners" / Montreal Model:

Antoine Boivin et al., Co-construire la santé en partenariat avec les patients et le public: historique, approche et impacts du "modèle de Montréal", available at <https://www.chairepartenariat.ca/wp-content/uploads/2019/07/Boivin-2017-Co-construire-la-sante%CC%81.pdf>

Karazivan P, Dumez V, Flora L, Pomey MP, Del Grande C, Ghadiri DP, Fernandez N, Jouet E, Las Vergnas O, Lebel P. The patient-as-partner approach in health care: a conceptual framework for a necessary transition. Acad Med. 2015;90(4):437-441. doi:10.1097/ACM.0000000000000603

Pomey MP, Flora L, Karazivan P, Dumez V, Lebel P, Vanier MC, Débarges B, Clavel N, Jouet É. Le « Montreal model » : enjeux du partenariat relationnel entre patients et professionnels de la santé. *Santé Publique*. 2015;27(Suppl 1):S41-S50. doi:10.3917/spub.150.0041

Bird M, Ouellet G, Whitmore C, Li L, Palacio DA, Whitehorne M, et al. Preparing for patient partnership: A scoping review of patient partner engagement and evaluation in research. *Health Expect*. 2020;23(3):523-539. doi:10.1111/hex.13039

Boivin A, Lehoux P, Burgers J, Grol R. What are the key ingredients for effective public involvement in health care improvement and policy decisions? A randomized trial process evaluation. *Milbank Q*. 2014;92(2):319-350. doi:10.1111/1468-0009.12060

Carman KL, Workman TA. Engaging patients and consumers in research evidence: Applying the conceptual model of patient and family engagement. *Patient Educ Couns*. 2017;100(1):25-29. doi:10.1016/j.pec.2016.07.009

Shippee ND, Domecq Garces JP, Prutsky Lopez GJ, Wang Z, Elraiyah TA, Nabhan M, et al. Patient and service user engagement in research: a systematic review and synthesized framework. *Health Expect*. 2015;18(5):1151-1166. doi:10.1111/hex.12090

## 2. Patient Partnership in Rare Disease Research

Velvin G, Hartman T, Bathen T. Patient involvement in rare diseases research: a scoping review of the literature and mixed method evaluation of Norwegian researchers' experiences and perceptions. *Orphanet J Rare Dis*. 2022;17(1):212. doi:10.1186/s13023-022-02357-y

European Joint Programme for Rare Diseases (EJP RD) / EURORDIS. Short Guide on Patient Partnerships in Rare Disease Research Projects - Basic, Pre-Clinical, Translational and Social Research. 2020/2025 edition. <https://www.ejprarediseases.org/wp-content/uploads/2020/08/SHORT-GUIDE-ON-PATIENT-PARTNER-SHIPS-IN-RARE-DISEASE-RESEARCH-PROJECTS.pdf>

## 3. Practical Frameworks and Use Cases for Patient Partners

Unité de soutien SSA Québec. 5 Ways Patient Partners Can Be Involved. 2024. <https://ssaquebec.ca/en/news/5-ways-patient-partners-can-be-involved/>

Centre of Excellence on Partnership with Patients and the Public (CEPPP). What is patient and public partnership? 7 must-read articles to better understand and deploy it. 2021. <https://ceppp.ca/en/news/what-is-patient-and-public-partnership-7-must-read-articles-to-better-understand-and-deploy-it/>

Abelson J, Humphrey A, Syrowatka A, Bidonde J, Judd M. Evaluating Patient, Family and Public Engagement in Health Services Improvement and System Redesign. *Healthc Q*. 2018;21(S1):61-67. doi:10.12927/hcq.2018.25636

## 4. Cooperative R&D Models and CMT-Focused Research Foundations

Burns J, Timmerman V, Mukherjee-Clavin B, Yiu EM, D'Antonio M, Laura M, Scherer SS, De Winter J. Charcot-Marie-Tooth disease and related neuropathies. *Nat Rev Dis Primers*. 2026;12(1):3. doi:10.1038/s41572-025-00008-3

#### 5. Additional Supporting References

Domecq JP, Prutsky G, Elraiyah T, Wang Z, Nabhan M, Shippee N, et al. Patient engagement in research: a systematic review. *BMC Health Serv Res*. 2014;14:89. doi:10.1186/1472-6963-14-89