

September 19, 2025

# Patient Readiness for Partnership in Research on inherited Neuromuscular Diseases (iNMD)

## Report of a Survey

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### Background

ECMTF and ECRA are striving to mobilize patients for partnership in research on inherited neuromuscular diseases (iNMD). "Patients as Partners" is the motto. It means that patients with their respective diseases are not just passive objects of medical care but can actively contribute to the progress of research and the development of therapies and drugs towards finding an adequate cure. This survey was addressed to patient advocacy groups, mainly in Europe, but also including the United States and others. The introduction to the survey reads as follows:

*Dear ECMTF members and other Patient Advocacy Groups,*

*The European CMT Federation (ECMTF) is conducting a brief survey to better understand the landscape of patient engagement in inherited neuromuscular diseases (iNMD) research. Our goal is to assess the readiness and interest of patients, as represented by your group, to move beyond being passive patients and into active partnership roles particularly in research projects and clinical trials.*

*For the purpose of this survey, "partnership" refers to active collaboration, such as surveys, research projects and clinical trials, helping to design studies, set research priorities, or serve on advisory boards.*

*Your insights are invaluable for helping researchers, sponsors, and policymakers create more meaningful and effective patient-centric research. The survey should take approximately 7 -10 minutes to complete. All responses will be aggregated and anonymized. The results of the survey will be presented and discussed at the 2nd European CMT Specialist Conference this October 23-25 in Antwerp.*

*Thank you for your time and contribution.*

Though the answers to the questions put are not based upon systematic surveys among affected patients but rather upon estimates of the management of the patient organizations concerned, we believe that they are meaningful in two respects:

First, the very fact that patient advocacy groups do exist and are active as self-help organizations striving towards relief of the symptoms, better management of the disease and, finally, the finding of a cure, proves that patients are not passive but taking already control of their own destiny.

Second, the answers in the survey provide us with a certain insight into how patients, through their organizations, have already developed strategies and priorities in pursuing their goal to improve their health condition, which are these strategies and priorities, and to what extent they are shared by the diverse organizations.

If it is true that only a small percentage – perhaps 5% - of the CMT-patients and patients affected by other iNMD are organised in patient advocacy groups, their membership and work is far from negligible. Patients' partnership in research is a condition of successful research, and there is no other legitimate reason for conducting research than helping the patients to get rid of – or at least alleviate – their disease. With their individual experience and natural history, but also as participants in clinical trials and in many other respects their commitment to partner with scientists, clinicians and other health professionals, thus, can make a difference.

This survey aims at providing a picture of where we stand today and what may be adequate strategies to mobilise patients as well as researchers recognizing and developing the opportunities and possibilities that a close partnership offers in each individual case for the benefit of all.

## **Methods: Survey design and distribution**

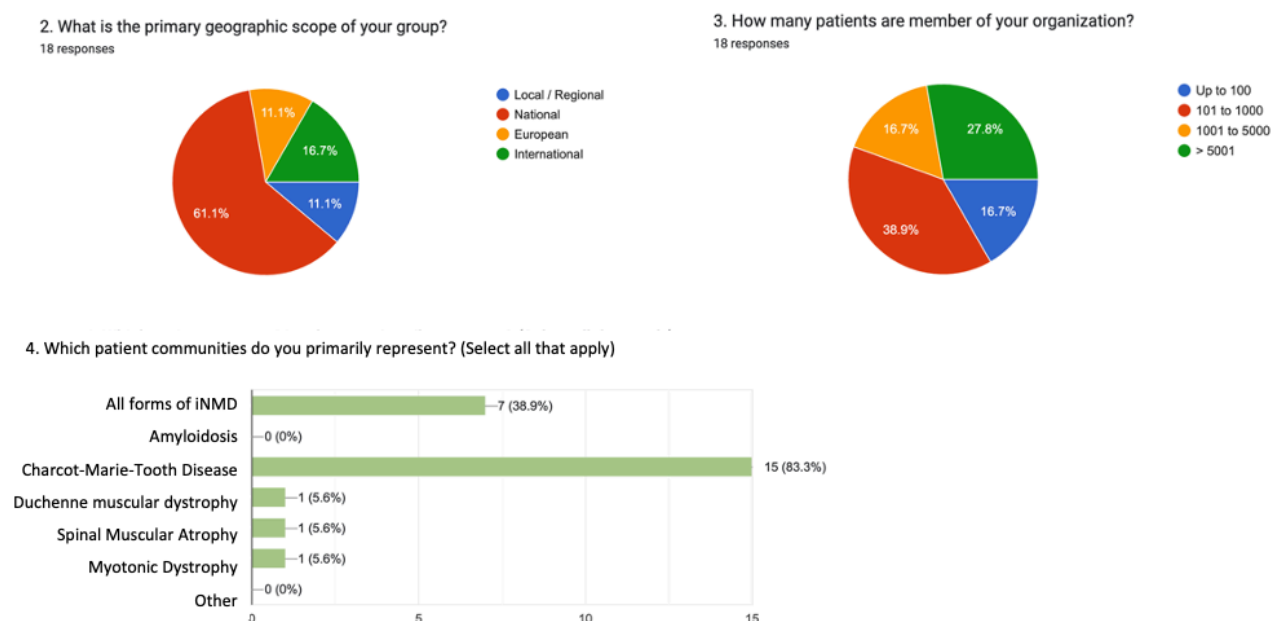
We designed a digital survey in preparation for the 2<sup>nd</sup> European CMT Specialist Conference in Antwerp (10/2025) in the context of the European CMT Research Association ECRA and in close collaboration with the European CMT Federation ECMTF and the University of Antwerp. We included five sections: the first section (questions 1 to 4) regard the description of the organisation, the second section (questions 5 and 6) is about participation of the organisation in clinical trials, the third section (questions 7 to 9) is about the demand among the patients members to the organisations, for information on research and trials, and the fourth section (questions 10 to 13) focuses on patients readiness for participation, effective participation and barriers to participation in clinical trials. The last and fifth section allowed for free text comments and recommendations. Most questions were posed as 0–10-point Likert-scales, with 0 points indicating negative and 10 points affirmative replies, unless stated otherwise in the question. The original questions are depicted in each figure with respective results plotted in the results section. The survey was conducted digitally using Google Forms between July and September 2025, with invitations sent via e-mail to around 80 addresses of patient organisations, their presidents or other officers and spokespersons from Europe and beyond, including from the United States.

## Results

18 participating organisations (22.5%) sent a reply, all replies included answers to all questions. Charts and tables showing the aggregated answers are depicted below. Diversities regarding the geographical scope, size, and focus of the organisations are not being considered. As we have promised anonymity, the names of the participating patient organizations (question 1 of the survey) are not published here.

### Section 1: Diversity of the organisations in scope, size, and focus

Regarding the geographic scope and the size of the organisations, those who replied were rather diverse under both aspects (Figure 1). 61% of the organisations replied to have a national scope, while only 16,7% said to have an international scope, and 11,1% were European and local/regional respectively. There is no genuine international patient organisation for iNMD and CMT and less than 30% of the organisations have an international scope, though the need for cooperation across the borders, if not worldwide, is evident. Also, the size of the organisations was quite diverse. Most of the organisations reported to have between 101 and 1000 members. 16,7% of the organisations have between 1000 and 5000 members, 16,7% have less than 100 members. 27,8% have more than 5000. The focus on CMT was most common, given that 38,9% replied to cover all forms of iNMD, and 83,3% to cover CMT. There was no reply for Amyloidosis and other iNMDs.



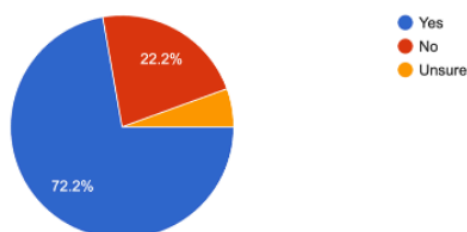
**Figure 1:** Pie charts and bar charts depicting the replies of patient organizations regarding diversity of the organisations in scope (question 2), size (question 3), and disease focus (question 3).

## Section 2: Activities of the organisations: Involvement in research or clinical trials

Almost three quarters of the organizations answered that they had been involved in research or clinical trials in the past 3 years, while less than one quarter did not. As can be seen in figure 2, question 6, funding research, promoting clinical trials and disseminating research results were the most common modalities of engagement (50%). Eight of 18 organisations signalled that they had a representative in a steering committee or advisory board of research projects.

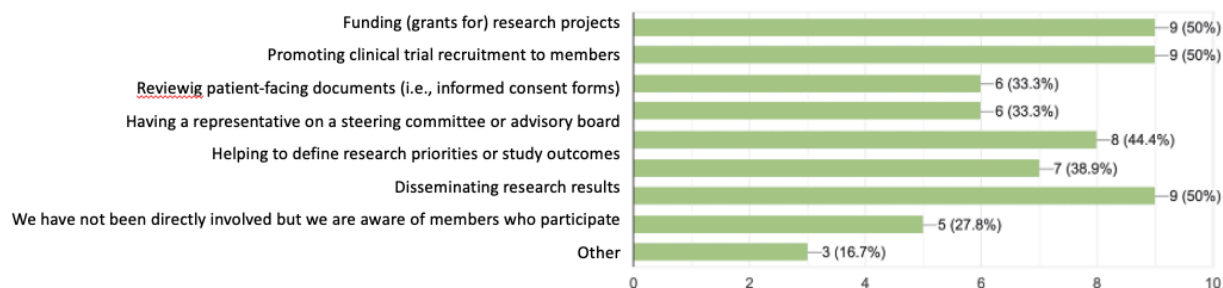
5. Has your group been involved in research or clinical trials in the past 3 years?

18 responses



6. In what capacity was your group involved? (Select all that apply)

18 responses



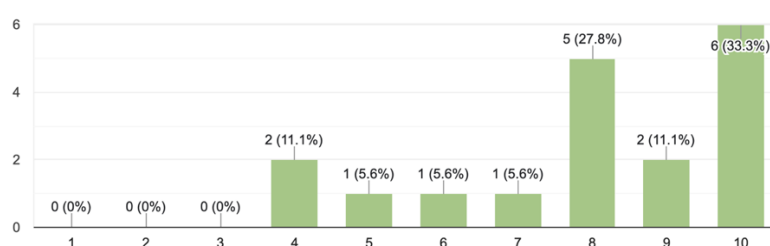
**Figure 2:** Pie charts and bar charts depicting the replies on activity of the organisations (question 5) and involvement (question 6) in research or clinical trials.

### Section 3: Information and the interface between patients and research

Questions regarding section three revealed that information on achievements in research, on new research projects and what academic institutions are undertaking, but also on clinical trials initiated by pharmaceutical companies, were high priorities in the tasks of patient organizations. More than 60% of the organisations stated a high and very high demand for information on new achievements and academic projects, for clinical trials the rate was even more than 65% (see figure 3 depicting questions 7, 8 and 9).

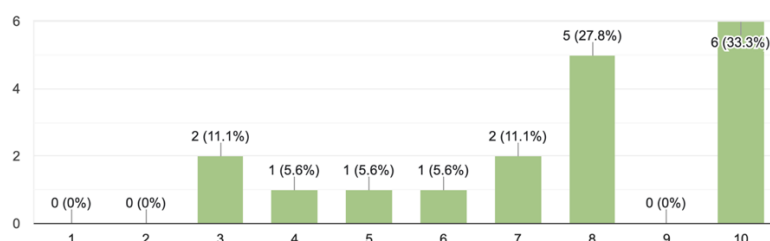
7. How would you rate the demand among your members for being informed on new achievements in INMD / CMT research?

18 responses



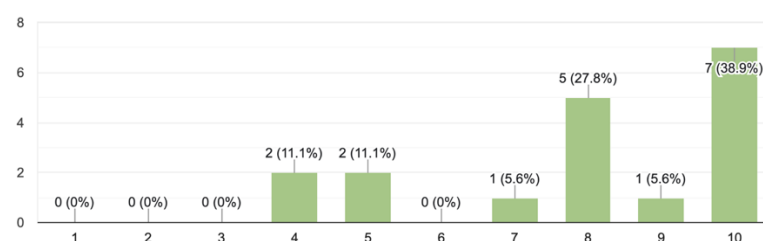
8. How would you rate the demand among your members for being informed on new research projects initiated by academic institutions?

18 responses



9. How would you rate the demand among your members for being informed on clinical trials, initiated by pharmaceutical companies?

18 responses



**Figure 3:** Bar plots representing replies on information and the interface between patients and research. X-axis depict Likert scale with 0 representing no demand and 10 very high demand.

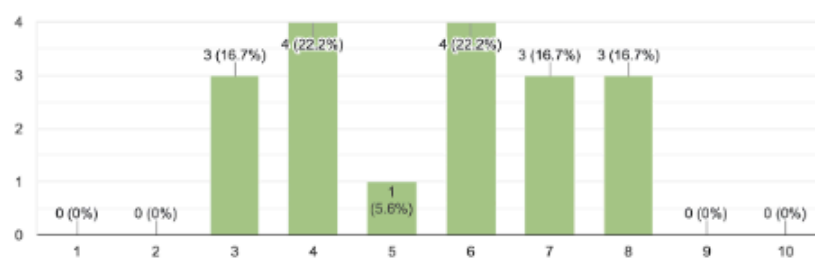
## Section 4: Patients' participation in research and clinical trials

In the next section, we asked for patients' awareness, interest, and willingness to participate in research and clinical trials (figure 4). Here, members' awareness of opportunities to participate in research was rated between medium and rather high. The willingness to participate in clinical trials was reported to be higher in comparison, with three patient organizations reporting a willingness of 100% (figure 4, question 10b). The interest in taking more responsibility was rated as rather poor, with most organizations reporting that the interest in active partnership roles was medium or lower for the patients they represent.

10. On behalf of the patients you represent, please answer the following questions:

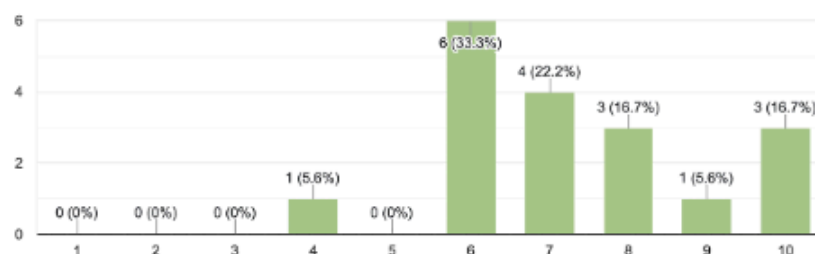
a) How would you rate your members' general awareness of opportunities to participate in research?

18 responses



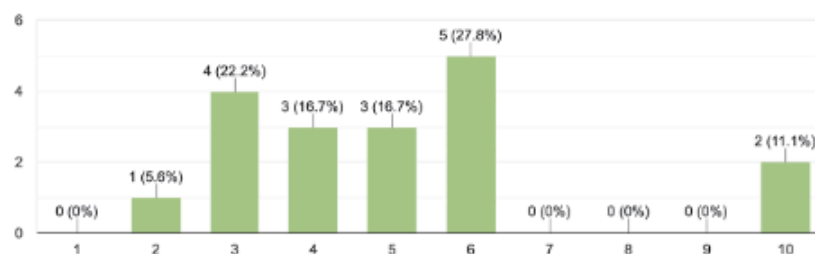
b) How would you rate your members' willingness to participate as subjects in a clinical trial?

18 responses



c). How would you rate your members' interest in taking on more active partnership roles, e.g., serving on advisory boards, co-designing studies?

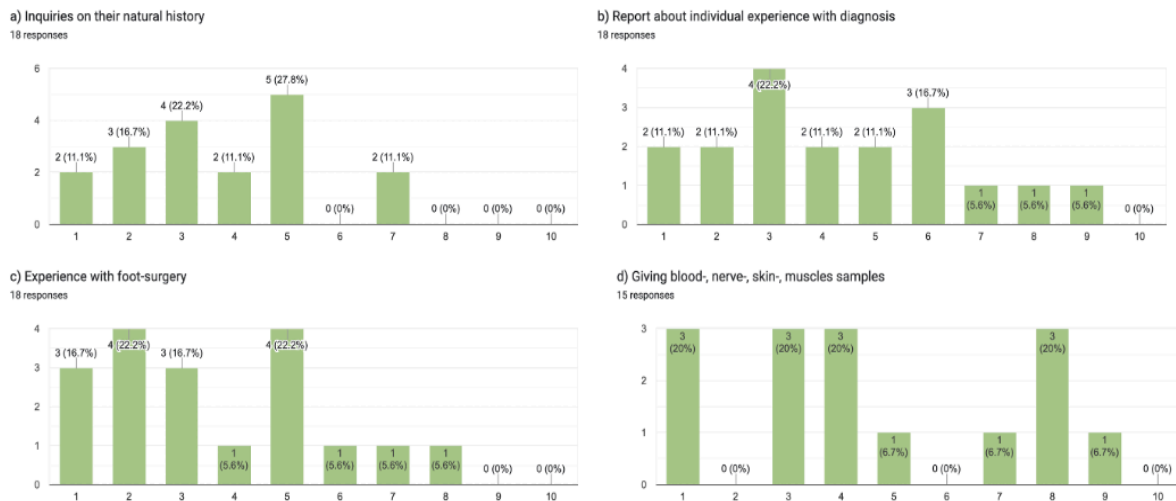
18 responses



**Figure 4:** Bar plots representing replies on patients' interest in participating in research and clinical trials as well as active roles of partnership. X-axes depict Likert scales with 0 representing no interest or participation and 10 very high interest or participation.

Similarly, organizations rated the effective participation of their members in research projects rather modestly: for inquiries on their national history, almost 90% of the organisations said that 50% or less of their members are participating; the picture is slightly more balanced regarding the experience with diagnoses and donation of blood-, nerve-, skin- and muscles samples, while half of the organisations indicated that 50% or less of their members had experience with foot surgery (figure 5, question 11 a-d).

11. To your knowledge, how many members of your organisation effectively participate(d) in research projects (1 = 0%, 10 = 100%), such as:



**Figure 5:** Bar plots representing replies on participation in research projects. X-axes depict Likert scales with 0 representing no participation and 10 representing 100% of patients participating.

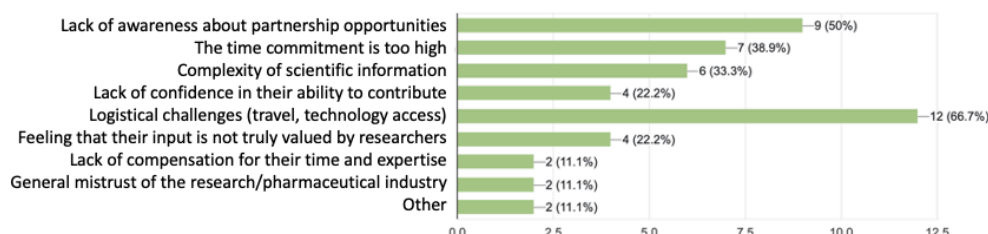
Answers further revealed that patients are often not aware of opportunities to partner with researchers or clinicians, or they hesitate to engage given the high time commitment, the complexity of the research topics and processes, or the logistical challenges such as typical mobility problems due to CMT, or issues with the access to the technology (figure 6, question 12). Organizations participating in this survey could choose 3 of 9 challenges. What is called logistical challenges got the highest rating (66,7%), followed by the lack of awareness (50%) and time commitment (38,9%). Lack of compensation, general mistrust and other reasons seemed to be of minor importance (11.1% each).

We next asked which methods patient organisations consider being the most adequate to increase readiness to partner with research (figure 6, question 13). Again, information was the priority: plain language summaries on research / partnership opportunities and results (61,1%). Here, funding to support patients' engagement, was mentioned second (55,6%). A dedicated platform to connect patients with researchers, and clear guidelines for how to engage with research teams were both chosen by one third of the

organisations participating in the survey. Opportunities to network were chosen by only 3 organisations.

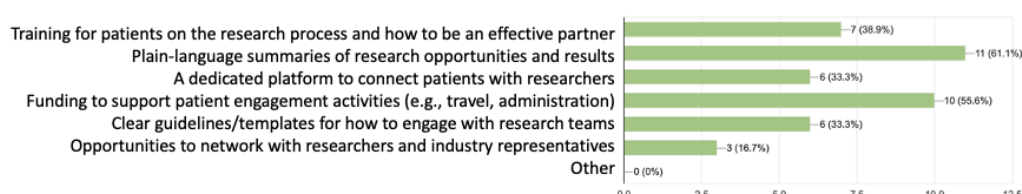
12. From your perspective, what are the biggest barriers preventing your members from engaging more deeply in research partnerships? (Select up to three)

18 responses



13. What would most help your organization and its members to become better research partners? (Select up to three)

18 responses



**Figure 6:** Bar plots representing replies on barriers to participation and partnership as well as potential opportunities to increase patients' participation in research.

## Section 5: Open comments

Finally, organizations were asked for open text comments about patient partnerships. Answers are depicted below (with minor edits from our side):

14. Is there anything else you would like to share about patient readiness for partnership in iNMD- or, in particular, CMT research?

- Getting in touch with the National Rare Disease Research Center, they should coordinate this research in the country and build relationships, not civil organizations.
- Essential to develop and validate outcome measures that are more sensitive to change.
- We must urgently move forward
- Patients need to know and need to be connected to experts to know that the investigation exists. People don't know about investigation
- I could not fill in where i Galerie the answer others: is asking questions and initializing study-designs live measuring gait on a foamy ground and analyzing the muscles which have to work different. And Studios about the effectiveness of trainings-therapy, electrotherapy, rhythm massage, music-therapy. One reason we do not participate more often is because we are not asked because our country is too little.



- Patients and families affected by CMT and other iNMDs are increasingly eager to move beyond being passive participants to becoming true partners in research. However, meaningful engagement requires support: educational resources to build research literacy, accessible communication about study goals and design, and mechanisms for feedback and co-creation. Many patients are ready and willing to contribute, but may lack the tools or confidence to fully participate unless those barriers are addressed. Additionally, cultural and language diversity, physical limitations, and digital access must be considered to ensure inclusive engagement. Strengthening collaboration between researchers and patient advocacy groups can help bridge these gaps and ensure that patient priorities shape the future of CMT research.
- Better information about the benefits of patient partnership in research, compensation for spending time on it.
- It wasn't possible to specify others if needed. We already have a dedicated platform to connect patients with researchers, and it is highly effective.
- In our experience, patients demand a lot (information, research...) but when they are asked to participate (for example a 1 minute video explaining how they live with CMT) the response is almost zero. Out of a group of 100-150 people, only 4 participated.
- Patients with CMT have heard for too long that treatments are coming. I believe that the trials in disease modifying technologies will become quickly fully subscribed. The natural history studies are less well-understood so there's greater hesitation. There seems to be reluctance to engage due to the lack of trust that their engagement is beneficial or that it can make an impact. Community members don't think that engagement will change their CMT experience and that it might not be worth the risk to help someone else.

## Discussion

Taken together, the survey shows that there is high interest of patients in research, but currently rather low participation or partnership, with major challenges regarding awareness of ongoing research, accessibility and logistics of research opportunities and trust as to the benefits of participation.

With only 18 replies to the invitation of around 80 patient advocacy groups, their leaders and organisations to participate in the survey, one may conclude that the readiness of patients to engage in partnership with science and clinical research is rather limited. Yet, this would be premature, since participation of an organisation in a survey is not the same as readiness of patients to

effectively partner in research. Also, the time frame allowed for organizations to answer fell within the summer holiday time, potentially limiting resources and available personal. With this in mind, the response to the initiative appears rather satisfactory. Regard needs to be given also to the fact, that with regard to GDPR organizations may not have exact information or may hesitate to give it.

The differences in size and the number of patients represented by each patient advocacy groups participating in the survey may be explained by the different size of the countries where each organisation is operating, but also with their diversity and different age. Also the experience concerning partnership with clinical or scientific research varies considerably. This may explain the differences in the numbers given by the organisations regarding awareness, willingness and effective participation of patients in research.

Comparing the assumed prevalence of CMT (1:2500) and the effective population of the respective countries with the numbers of members of each association, CMT/iNMD it appears that patients are either reluctant to engage in patient advocacy groups or are even not aware opportunities offered by such groups. Challenges of the disease like fatigue or mobility restrictions that slow down everyday activities, may also limit resources for commitment beyond daily activities. Yet, there seems to be a clear need for awareness rising campaigns and mobilizing patients to act together and partner with research if they wish to see progress in finding a cure for their disease.

Research and participation in clinical trials seem to be of high relevance both, for the organizations and for their members. As shown in figure 2 question 5 almost three quarters of the patient advocacy groups indicate that they were active in these fields in the last 3 years. Half of the organizations specified that they were active in funding research projects and promoting clinical trial recruitment (figure 2 question 6).

Since most of the patient organizations having participated to the survey represent CMT patients, it is fair to say that this survey does reflect the situation for CMT. The diversity of the groups in scope and size is remarkable, yet it does not seem to be relevant for the interpretation of the results of the survey.

Patients join advocacy groups for several purposes: organising self-help, getting advice and exchanging experience among themselves, giving patients a voice in politics and public sphere, and promoting research. Information on achievements in research, on new research projects and what academic institutions are undertaking, but also on clinical trials initiated by pharmaceutical companies are, thus, high priorities of the organizations.

In contrast, members' *awareness* of opportunities to participate in research was reported as rated rather low in comparison, which could explain the high interest in being informed. Instead, the *willingness* to participate in clinical trials was comparably high. Potential reasons for this gap as well as for patients' rather low readiness to take active roles include a lack of access to research, logistical challenges, and a fear of or inability to provide the expected time commitment. When patients participate in research, this includes mostly natural history and donation of tissue samples. Less patients had experience with foot surgery – which may be explained by the fact that foot deformation that needs surgery is a symptom that occurs only to a part of the CMT patients. In contrast, the other forms of participation are generally applicable to all CMT and iNMD patients.

Barriers of participation included mainly logistical challenges and lack of awareness, as prior results also suggested. Potential reasons may be that the more heavily people are disabled, the less they are ready to move to the clinics or laboratories, to meetings or to their specialists or even to their local doctors. A possible remedy could be to make study visits more comfortable. Combining a study visit with routine visits (which is already done) to avoid extra commute times, implementing calls and virtual visits, and conducting all diagnostic tests in one day would probably help. Digital devices for communication and digital care may also be of help on these challenges if they are easy enough to use. They may also reduce the time burden.

Information on the opportunities to partner is another key condition for patients' readiness for partnership. More systematic and plain language information and education on CMT, its genetic causes and methods to cope with it would reduce the complexity of the issues patients are confronted with, reduce the feeling that their contribution would be of no value to research, and so encourage patients to engage in partnership with science.

All the methods mentioned are closely related to each other. If cooperation and partnership of patient organisations with the researchers/clinicians and their organisations does already exist, it is nevertheless fragmented so far and not a common practice. It should become the standard, part of the system. The same applies for individuals: An organization referred to in one of the comments rightly calls for considering patients not as passengers, but as “co-pilots.” Where personal contact may be too burdensome to organize, the digital tools may be highly effective also for connecting patients with researchers. This may help to solve some of the current roadblocks, and serve as an example to move forward.

## Conclusions

The findings of this survey on patient readiness for partnership in research from the point of view of patient organization representatives collectively underline the high need for better information of patients on the impact they can have on research. Key conclusions are:

- Clear and reliable information and education on what the disease really is and what approaches are taken in the research community, including needs and steps for new therapy approval by EMA and FDA, is a top priority.
- Dedicated (digital) platforms allowing patients and researchers to meet, connect and explore opportunities and procedures of participation – and experience partnership – seem to be highly effective. New technologies and easily accessible tools for digital care may bridge geographical gaps.
- Patient advocacy groups play an important role as interface among patients and researchers/clinicians. Their partnership with researcher's associations promise considerable advances in research for the benefit of patients.

To promote such improvements and move forward in better including patients in current and future research, we are confident that the partnership of ECMTF with ECRA, and their joint conferences like the 2<sup>nd</sup> ECMTSC in Antwerp 2025 will be a way ahead.

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## Readings and references:

- For an early initiative: Antoine Boivin et al., Co-construire la santé en partenariat avec les patients et le public : historique, approche et impacts du « modèle de Montréal », <https://www.chairepartenariat.ca/wp-content/uploads/2019/07/Boivin-2017-Co-construire-la-santé.pdf>.
- For a practical initiative: CMTA, Patients as Partner in Research. Real People. Real Impact, <https://cmtausa.org/patients-as-partners-in-research/>.
- Quote: „In our case, patients are not passengers in their own diagnostic Odyssey, but co-pilots“..., Institute of Genomic Medicine and Rare Disorders, Semmelweis University, Faculty of Medicine, <https://semmelweis.hu/genomikai-medicina/en/about-us/>.