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Wellbeing measurement among adults with Charcot-Marie-Tooth disease

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ABSTRACT

Background and aims: Wellbeing research among individuals with Charcot-Marie-Tooth disease (CMT) is limited. The goal of the current study is to characterize the challenges experienced by adults with CMT that researchers may miss by utilizing typical strategies to capture mental health and wellbeing.
Methods: In 2021, we recruited 288 US adults with CMT (M_{Age} = 60 years, 59% Female, 93% White) to take part in an online survey. Participants were presented with validated wellbeing and health measures, in addition to newly created measures specific to CMT, and then asked to provide free-response feedback about aspects of their lived experiences missing from these measures. Thematic analysis were applied to the free-response data to identify CMT-related difficulties that may impact wellbeing among adults with CMT.
Results: Results identified nine CMT-related challenges that are important to individuals with CMT and are often left out of wellbeing assessments in this population. The three most common themes identified were anxiety or worry, coping with loss of function or progression, and difficulty finding knowledgeable medical providers. Further, some aspects of the CMT experience may be comorbid.
Interpretation: These results can be used to inform future research and clinical practice in the CMT population.

Research on wellbeing among people with disabilities has gained popularity in recent years, due in part to the increased recognition of health as more than simply the absence of diagnoses. However, wellbeing measures rarely take into account the perspective of those with disabilities. Commonly employed approaches to assessing wellbeing may lead to inaccurate or incomplete measures among individuals with disabilities for a couple different reasons. First, measures may fail to comprehensively capture the domains of importance to disabled participants.[1] One area in which this can be seen is in quality-of-life measures. Quality of life, a component of wellbeing, is an individualized construct, insofar that the domains that determine the quality of one's life vary across individuals.[2] However, many quality-of-life measures only assess a limited selection of domains, meaning many domains that individuals perceive as essential for quality of life may not even be assessed. This is especially important for people with disabilities, as research suggests a wide variety of domains that may greatly impact quality of life in clinical populations that are not captured through most quality-of-life measures.[3,4]

Second, many wellbeing measures do not assess the relative importance of each construct or domain.[1] For example, physical functioning is a common domain in quality-of-life measures. However, physical functioning measures will almost automatically consider one as in "low status" or of "low QoL" if they have a physical disability. This is true even if the individual does not place much value on their physical functioning or has adapted to do the things they need and want to do in life. For example, an item in the Official Short Form-36 (SF-36) measure;[5] developed at RAND as part of the Medical Outcomes Study), a commonly employed measure of health-related QoL, asks participants, "How much does your health now limit you in walking more than a mile?" Individuals with physical disabilities who struggle or are unable to complete this task would be deemed to have a lower health-related QoL by this measure. However, as discussed by Andresen and Meyers,[6] these individuals may be able to traverse the same distance using a wheelchair or other assistive device. Therefore, the inability to walk more than a mile may not actually impact QoL for many individuals. This can also be seen in other wellbeing measurements such as activity engagement. Activity engagement measures often list activities and ask participants which ones they engage in and how often. However, many of these measures can be viewed as ableist in nature as they contain items that may be less relevant to individuals with disabilities. For example, walking is a common activity listed on activity engagement measures (e.g.,[7]); however, this may not be applicable to some wheelchair users. Activity engagement measures also often ask about attending various public places such as theaters and museums (e.g.,[8]).

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However, many of these venues may not be accessible to people with disabilities; therefore, the opportunities for disabled people to engage in these activities may be limited. Because these measures contain many items that may not be applicable or valued for people with disabilities, these individuals may receive lower engagement scores, even if they are highly engaged in other activities not captured by these measures. Therefore, it is important for researchers to consider the perspectives of people with disabilities when designing measures to assess wellbeing-related constructs in this population.

Although the issues listed above hold in general for research with people with disabilities, it is also clear that disabled people are a broad, heterogeneous collective who face differing life circumstances and consequences from their disability status. As such, another concern is that by focusing on disability status broadly, researchers may miss the nuances and unique factors that impact wellbeing for specific groups.[1,9] One population that has been underrepresented in wellbeing research is Charcot-Marie-Tooth disease (CMT), an inherited neuromuscular disorder affecting over three million people worldwide.[10] CMT affects the peripheral nervous system and leads to progressive muscle weakness, fatigue, pain, decreased balance, and gradual loss of physical functioning.[11] Onset is typically in the first two decades of life, although this can vary greatly.[10]

Although limited, prior studies suggest that individuals with CMT may experience lower wellbeing and mental health than individuals without CMT, although results are mixed. [12] One factor that may greatly impact wellbeing and health for individuals with CMT are healthcare transitions. Research in other clinical populations suggests that adolescent to adult healthcare transitions are important for promoting treatment adherence and health outcomes.[13,14] Most research on healthcare transitions in general have focused on transitions to adult care during adolescence and young adulthood. However, as pointed out in the editor's opening commentary for *Health Care Transitions*, in order to fully understand the consequences of long-term health care transition planning, it may be important to go past emerging adulthood when studying transitions.[15] However, little work has examined healthcare transitions among adults with CMT.

Considering that CMT may lead to worse wellbeing, and samples with CMT have been underrepresented in psychological research, [11,12,16,17] work is needed that recognizes the unique experience of CMT and captures important factors such as the unpredictable progressive nature of CMT, its hereditary pattern, and rare classification that may further impact wellbeing in this population. In addition, since little research has examined healthcare transitions among people with CMT, work is needed to examine to what extent aspects of adolescent to adult healthcare transitions may impact wellbeing among this population. The current study will address this using an open-ended survey approach among a nationwide sample of U.S. adults with CMT. This open-ended approach was chosen to encourage responses that may not have been captured through use of quantitative questionnaires due to the researcher-imposed framework and more limited scope. The goal of this study was to characterize what psychological and social challenges researchers may miss by solely utilizing typical strategies to capture wellbeing among people with CMT, helping to catalyze future research.

1. Current study

Our sample of adults with CMT first completed several traditionally employed quantitative measures (see[18]). Participants were then asked for their feedback about what was missing from our study. Specifically, they were asked "Are there any other obstacles or challenges you face due to your CMT that you feel were not addressed in this survey? If so, please elaborate" and "Is there anything else you would like to note about your experiences living with CMT?".

2. Methods and materials

Participant recruitment for this study was conducted in collaboration with the Charcot-Marie-Tooth Association (CMTA). After discussing this project with the CMTA, the organization agreed to provide recruitment assistance on this project. Participants were recruited in 2021 via social media posts on the CMTA's Twitter, Instagram, and Facebook Group pages and email blasts to the CMTA's database. No compensation was provided to participants; however, upon completion of the survey, participants were given the option to be entered into a raffle for one of two $50 gift cards.

2.1. Materials/measures

After clicking on the survey link, participants were shown a consent information sheet and asked, "Do you consent to participate in the study?" Only participants who selected "Yes" were included in the study. Participants were first asked to complete 12 quantitative measures that were chosen to capture commonly assessed constructs in disability psychology research: discrimination experiences, CMT-related anxiety, avoidance due to CMT-related anxiety, self-efficacy, adaptive device use, adaptive device helpfulness, CMT/disability community perceived support, involvement in the CMT/disability community, perceived social support, sense of purpose, life satisfaction, and health-related quality of life (HRQoL) (see[18] for more details). After completing these quantitative measures, participants were asked to answer two open-ended questions regarding the challenges they face due to their CMT: (1) "Are there any other obstacles or challenges you face due to your CMT that you feel were not addressed in this survey? If so, please elaborate." and (2) "Is there anything else you would like to note about your experiences living with CMT?".

2.2. Analytic method

First, the authors compiled a list of expected qualitative themes based on past disability studies as well as the first author's experiences running a CMT support group. Second, the first author read through the open-ended responses and revised the list of themes to include additional emergent themes. The first author then analyzed each participant response for alignment with the emerging themes. Next, two co-authors independently analyzed data from 30 participants utilizing the same coding as the first author. All three analyses were compared; interrater reliability ranged from .67 to 1 across all themes. Discrepancies were addressed until all three authors were in agreement. Frequencies were then calculated for each of the themes. Chi-squared analyses were also conducted between each of the themes to explore whether individuals reporting certain themes were more likely to also report others. Put differently, these tests examine whether participants who viewed one construct or experience as important tended to discuss another theme as well.

2.3. Transparency and openness

This research was reviewed and considered exempt by the Washington University in St. Louis Institutional Review Board due to the anonymity of participants and lack of experimental manipulation. These data were collected as part of a study that also administered quantitative measures related to wellbeing and health. The design and analyses for this overall study were preregistered; see https://osf.io/hc384/?view_only=c6dfeca870e3402cbee9cb4121f14965. The first author is a member of the CMT community and a volunteer for the CMTA.

3. Results

3.1. Participants

Participants included 315 adults (M_{Age} = 59.26, 59.27% Female, 91.84% White) who took part in an approximately 10–20-minute online survey. Twenty-seven participants failed to meet eligibility criteria (less than 18 years of age, no CMT diagnosis, or lived outside of the US) or did not complete all eligibility items and were excluded from the analytic sample. Our final sample included 288 US adults with CMT (M_{Age} = 59.57 years, 59.30% Female, 92.70% White). A little under half of our sample (45.83%) did not report anything missing from our study.

3.2. Description

As shown in Table 1, nine challenges of having CMT emerged from the participants' open-ended responses. Of these nine themes, "Coping with loss of function or progression", "Anxiety or worry", "Difficulty finding knowledgeable medical providers", and "Lack of understanding from others" were most frequently reported. In contrast, "Feelings of loneliness" and "Systemic discrimination" were least common among our sample. Three unanticipated themes also emerged from our data: difficulty finding knowledgeable medical providers, feelings of self-consciousness surrounding CMT, and challenges of having comorbid mental or physical health conditions.

3.3. Most common themes

3.3.1. Anxiety or worry

Almost a quarter of participants who provided open ended responses (21.15%) reported experiencing anxiety or worry related to their CMT. The most commonly reported source of anxiety or worry was future disease progression (27.27%). Some prototypic examples from the participants included:

> "I am scared for my future in the sense of how much worse it's going to get and if I will ever be able to have a family."

> "By far the worst is the fear of what is to come and how much it will slow me down in my older years."

> "I worry a lot about where I will be able to live safely as I age and as my condition progresses."

Another commonly reported source of worry or anxiety was passing on CMT to one's offspring. The distinguishing feature in these responses was that individuals mentioned concerns regarding heritability and the concomitant guilt. Responses included:

> "I worry constantly that my children have inherited CMT."

> "The guilt of passing this disease on to my child(ren). It was a very real fear when discussing children with my spouse."

Other participants mentioned difficulties with functioning or meeting expectations in the workplace as a source of anxiety.

> "My hands are progressing with CMT quickly and I use them to do my job (write/type on the computer). This is a source of anxiety. Additionally, I am a professional and often cannot fit my AFOS (ankle foot orthoses) into appropriate shoes for my attire, and this also causes anxiety."

> "[I experience] fear and anxiety because of reduced job opportunities and the ability to make a living due to the loss of dexterity and coordination."

Some participants stated that they often worried about their ability to navigate their everyday lives due to their CMT.

Table 1
Emerging themes frequencies and definitions.

Theme	% Agreement	% Occurred in Open Ended Responses	Definition
Anxiety/worry	86.67	21.15	The participant describes feeling worried or anxious about CMT-related things.
Coping with loss of function or progression	66.67	19.87	The participant describes thoughts and behaviors they experience after losing function or realizing their CMT has progressed.
Difficulty finding knowledgeable medical providers	90.00	14.74	Participant describes having trouble finding care providers (e.g., physicians, physical therapists) who possess current, comprehensive, and correct knowledge about CMT.
Lack of understanding from others	86.67	12.18	The participant describes feeling that others do not understand their experiences with CMT.
Difficulty obtaining or using medical services or devices	90.00	10.90	The participant describes having difficulty obtaining needed medical devices or services (e.g., due to finances or lack of availability) or difficulty using medical devices (e.g., due to their visibility or need for accommodation).
Feelings of self-consciousness surrounding CMT	96.67	8.97	The participant describes feeling embarrassed, ashamed, or fearful of judgment from others due to their CMT.
Challenges of having comorbid mental or physical health conditions	93.33	7.05	Participant describes the interaction between CMT and their other mental or physical health diagnoses and the challenges they face due to these interactions.
Feelings of loneliness	100.00	6.41	The participant describes feeling alone in their struggles with CMT and/or that they do not have anyone to talk to about CMT-related topics.
Systemic discrimination (accommodations, inaccessibility, etc.)	96.67	5.77	Participants describes experiencing discrimination at a systemic level (e.g., having difficulty navigating public buildings, getting a job, securing work/school accommodations).

Description: This table displays the nine CMT-related challenges that were identified as important and impactful among individuals with CMT.

> "[It's] scary getting locked in public and private bathrooms since [my] hands and fingers [are] too weak to turn [the] lock to unlock. Even car doors at times are tough to open. Not fun. [It's] scary getting locked in somewhere due to not having strength in [my] hands."

> "[I experience] anxiety with driving daily or walking and my braces breaking."

3.3.2. Coping with loss of function or progression

About a fifth of participants (19.87%) who responded to the open-ended questions reported coping with CMT progression and resulting loss of function as a significant challenge. For instance, multiple participants described that giving up activities or occupations they previously enjoyed due to disease progression was especially difficult:

> "I was a RN until I was 63. I became more clumsy and my hands weaker and I was anxious about work. I retired early which was hard as I loved being a nurse."

> "I like to tell children who have an interest in my 'boots' and wheelchair, I have gone through resignation of independence, physical activities like dance and swimming. I was family caregiver. I fell a lot but can no longer get myself up."

> "I feel there is a strong need for increased mental health services for persons with CMT, or any physical disability, to help cope with the secondary depression, shame, and anxiety that frequently accompanies the loss of functioning."

Other participants reported trying to cope with loss of functioning by focusing instead on activities they can do.

> "Having CMT is depressing, but I try to concentrate on the things I can still do and consider myself lucky that I'm a writer so I can continue doing at least that activity if I can't do the things I loved doing in the past–e.g., traveling, walking in nature, gardening, playing tennis, camping."

3.3.3. Difficulty finding knowledgeable medical providers

Many participants (14.74%) reported that finding medical providers who are knowledgeable about CMT is one of the biggest challenges in living with CMT. Many reported that they felt like they had to educate their providers on CMT.

> "We DESPERATELY, DESPERATELY Need LOCAL access to more medical PROFESSIONALS that have both Current and Correct Knowledge of CMT, and its challenges and latest treatments. The people we need to count on the very most to help us, don't understand or have any knowledge about CMT, or if they do recall something it's nearly always antiquated."

> "The biggest obstacle I have faced in this journey is the overall ignorance of CMT. This is common in the general population, but it is astounding how little the medical community knows of CMT, let alone knows enough about it to provide assistance."

In addition to lacking knowledge of the biological aspect of CMT, participants also found that physicians lacked knowledge on secondary psychological aspects of the disease as well as of tools to help patients adapt to decreased physical functioning.

> "[It's difficult] finding health care providers that are knowledgeable about CMT, not just the scientific literature, but coping and hacks to ease life."

3.3.4. Lack of understanding from others

Some participants (12.18%) reported that a lack of understanding from others was one of the biggest challenges of living with CMT. Since CMT can sometimes be an invisible disability, some participants experienced judgmental or dismissive reactions from others due to CMT-related struggles or accommodations.

> "I get treated differently because this is an invisible disability and I seem normal to people. Therefore some co-workers treat me like I'm lazy."

> "I don't look disabled so my peers have difficulty understanding the extreme fatigue I get from physical activities such as hiking. My muscles are weak but look normal. And if I'm on my feet for many hours or traveling, I require a day to recover."

> "Since it is an invisible disability usually there are a lot of mean looks or comments when using handicap placard or sitting down on a crowded bus."

Other participants reported a general lack of awareness of CMT and understanding of their CMT-related experiences among people around them.

> "My biggest obstacle in dealing with my CMT is trying to educate people all the time to what it is…It gets frustrating."

> "It feels very lonely. Nobody in my life gets it."

3.4. Chi-squared analyses

First, as shown in Table 2, results indicate that those who experienced difficulty coping with loss of function or progression were more likely to also report challenges of having comorbid mental or physical health conditions, $X^2(156) = 6.75$, $p < .01$. Second, participants who reported feelings of self-consciousness surrounding CMT were more likely to also experience feelings of loneliness, $X^2(156) = 8.86$, $p < .01$. Third, individuals who reported a lack of understanding from others were more likely to experience feelings of loneliness, $X^2(156) = 5.20$, $p < .05$, and difficulty finding knowledgeable medical providers, $X^2(156) = 6.52$, $p < .05$. All other chi squared tests conducted were non-significant.

4. Discussion

This study was one of the first to qualitatively examine challenges associated with CMT from the perspective of individuals with CMT. Past research on wellbeing among people with CMT is scarce and wellbeing measures often do not take into account the perspective of those with CMT or disabilities in general. Our results suggested that, while individuals with CMT are fairly satisfied with traditional wellbeing measures, there are important aspects of their lived experience with CMT that are not fully captured by these measures. Specifically, this work identified and characterized nine CMT-related challenges that are important and impactful to individuals with CMT. Anxiety/worry, coping with CMT progression, and difficulty finding knowledgeable medical providers were the most frequently reported challenges among our sample while feelings of loneliness and systemic discrimination were reported least frequently. Further, our results suggest that some aspects of the CMT experience may be comorbid, insofar that people who mentioned a certain theme as a critical need for assessment also tended to mention additional themes. The following sections provide a further review of the findings of the study, their implications, and limitations.

4.1. Implications for measurement

This study provides valuable implications for wellbeing measurement among adults with CMT. First, just under half our sample did not list any missing aspects of our quantitative survey. This finding provides some promising news for research in the field, insofar that it suggests that traditional wellbeing measures adequately captured their lived experiences, and they are useful for assessing components of wellbeing in people with CMT. Second, our results also indicate that these traditional measures do fail to capture many CMT-related challenges that are important to individuals with CMT and their wellbeing. For instance, worry/anxiety, feeling a lack of understanding from others, and coping with disease progression were three of the most frequently reported challenges in our sample, which is noteworthy given past research linking these constructs to wellbeing.[18–20] Therefore, these areas merit attention in future measurement development. Third, our results found

Table 2
Chi-Squared Statistic for the nine themes identified.

	Anxiety/ worry	Coping with loss of function or progression	Difficulty finding knowledgeable medical providers	Lack of understanding from others	Difficulty obtaining or using medical services or devices	Feelings of self-consciousness surrounding CMT	Challenges of having comorbid mental or physical health conditions	Feelings of loneliness	Systemic discrimination
Anxiety/worry	-	2.09	0.12	0.00	0.48	1.11	0.81	0.09	1.80
Coping with loss of function or progression		-	0.37	0.61	0.32	1.45	6.75**	1.54	0.00
Difficulty finding knowledgeable medical providers			-	6.52**	0.52	1.53	0.01	0.00	0.00
Lack of understanding from others				-	0.00	2.36	0.64	5.20*	2.17
Difficulty obtaining or using medical services or devices					-	0.00	0.49	0.00	0.33
Feelings of self-consciousness surrounding CMT						-	0.31	8.86**	0.69
Challenges of having comorbid mental or physical health conditions							-	1.03	0.03
Feelings of loneliness								-	1.67
Systemic discrimination									-

Note. $^{**}p < .01$. $^{*}p < .05$.

that individuals reporting certain themes were more likely to also report others. Future research on wellbeing among individuals with CMT may benefit from the development and use of multifaceted measures of wellbeing that consider relevant CMT-related challenges identified in the current study in addition to traditional wellbeing constructs.

4.2. Implications for clinical practice

Although our study focused on measurement, the qualitative responses provided rich feedback valuable to better understanding healthcare transitions and clinical practice among individuals with CMT. First, one of the most common challenges reported by our sample was difficulty finding knowledgeable medical providers. This point suggests that many physicians, physical therapists, orthotists, and other care professionals may not receive adequate training on CMT. This finding aligns with prior research that found that patients with rare diseases often struggle to find medical professionals knowledgeable about their diagnosis, and in turn, they are forced to become experts in their disease in order to educate their doctors.[21] Prior research with non-CMT clinical samples suggests that lack of knowledge among medical providers may result in lower quality care and/or communication difficulties,[21,22] both of which may negatively impact well-being.[23] and discourage individuals from seeking future medical care.[24] Avoiding health care may lead to worse health outcomes down the road. More opportunities for medical professionals to learn updated information about CMT could help optimize care and wellbeing for people with CMT.

The finding that people with CMT often struggle to find knowledgeable medical providers maps onto previous work identifying adolescent to adult healthcare transition barriers, including a lack of available adult providers and a lack of providers with training in congenital conditions.[25] Somewhat surprisingly, participants did not specifically discuss the transition from adolescence-to-adulthood as a primary theme. One possibility is that participants had forgotten the difficulties inherent during that time, and future research should consider collecting samples younger in age to examine whether health care transition obstacles were forgotten or simply not significant enough barriers to merit attention. Another possibility speaks to the unique nature of CMT, given the variability in the age of symptom onset among individuals with CMT. While the general population often undergoes impactful healthcare transitions during adolescence and young adulthood, these timelines may vary greatly for CMT-specific healthcare. For individuals with CMT, healthcare transitions (e.g., seeing a new provider, seeking out a new kind of therapy or medical equipment) likely occur during the diagnosis process and during periods of disease progression or new loss of function. Since CMT symptoms may onset or progress at any point in an individual's life,[10] this population may not have a specific healthcare transitions period as it likely varies greatly across individuals with CMT. Therefore, individuals with CMT may have additional healthcare transition needs from the ones identified in the previously mentioned review paper. Future research is needed to identify specific healthcare transition and clinical practice concerns among individuals with CMT across the lifespan in order to help promote wellbeing and health for individuals with CMT.

Second, many challenges reported by individuals with CMT were psychosocial in nature. Because CMT is largely viewed as a physical health condition among medical professionals, standard assessment is often limited to physical factors such as strength and balance. In addition, treatment recommendations are often aimed at improving or maintaining physical functioning.[26] While those assessments and

interventions are extremely valuable, our results suggest that some of the biggest obstacles faced by individuals with CMT are psychological or social difficulties. This finding is in alignment with additional work with the current sample showing associations between psychosocial factors and wellbeing among individuals with CMT.[18] For example, our results suggest that many individuals with CMT experience anxiety about a variety of CMT-related factors and have difficulty coping with progression of their CMT. While these challenges are likely related to decreased physical functioning, regular assessment for these challenges at routine care appointments could allow doctors to refer patients who are struggling for psychotherapy or other interventions. Early intervention on these difficulties may be extremely beneficial for helping individuals better cope with these challenges and promoting wellbeing among individuals CMT.

4.3. Implications for understanding the CMT experience

Our results help further our understanding of the lived experience of individuals with CMT in a couple different ways. First, these results help to characterize the challenges that are impactful and important to individuals with CMT. Of the emergent themes, anxiety/worry and coping with loss of function or progression were reported most frequently. This finding suggests that these challenges are frequently experienced by individuals with CMT and likely impact wellbeing. While little work has examined these challenges among individuals with CMT, these findings align with prior work showing that individuals with similar physical disabilities often experience higher rates of anxiety, [27,28] and that loss of physical function may be associated with decreased quality of life. [29] Moreover, it is noteworthy that emergent themes focused on risk factors for negative psychological wellbeing, rather than positive aspects or resilience factors missing from past research.

Second, that said, the results also suggest that a deficit perspective is insufficient for understanding the CMT experience. While our sample did report psychological wellbeing issues, they did so at a much lower frequency than anticipated. In fact, roughly half of participants did not report any additional wellbeing related issues missing from past quantitative batteries. In addition, because previous research on disability has largely been conducted through a deficit perspective, the themes compiled based on our initial review of the literature were largely focused on deficits. Several of these themes though were not evidenced in our data. Paired with the low frequency with which participants reported psychological wellbeing issues, these findings suggest that the CMT experience cannot be fully captured by a deficit perspective. Instead, more research that avoids a deficit perspective is needed to more fully understand the experiences of individuals with CMT. This avoidance of a deficit perspective is also aligned with calls from disability activists for a movement towards the social model of disability, which views many of the limitations and participation barriers faced by disabled individuals as a direct result of inaccessible environments rather than of the impairment itself.[30] For example, rather than viewing CMT as the potential cause of lower wellbeing, research assessing the role of the physical and social environment on wellbeing among people with CMT may be beneficial for avoiding a deficit perspective. In addition, measures such as disability identity[31] and disability pride,[32] which view disability through an identity rather than a deficit lens, could be valuable for future wellbeing research among individuals with disabilities.

4.4. Limitations

Although this study provides valuable findings for future research with the CMT community, there are limitations that should be considered. First, this study was cross-sectional; therefore, a single assessment may be biased toward what someone was dealing with that day, and people may have failed to remember other relevant issues. Second, since this was an online survey, we were not able to ask follow-up questions or clarify participant responses. Third, since this study was conducted in the middle of a global pandemic, participants may have faced unique challenges that could have impacted their usual resources and wellbeing. However, it is worth noting that specific mentions of COVID-19 were very minimal in our data. Fourth, since participants reported on their own wellbeing and experiences, future research examining friend and family member reports may be useful for better understanding what challenges they perceive as impactful for their loved ones.

4.5. Constraints on generality

While we attempted to increase diversity in our sample by recruiting nationwide and utilizing multiple recruitment platforms, limiting factors within our sample demographics may impact the generalizability of these results. First, the majority of the sample was middle-to-older adults; therefore, it is unclear whether these findings are applicable to younger individuals with CMT. Second, the majority of our sample self-reported income in the middle to high brackets. Because SES influences access to medical care, adaptive devices, and other resources, research is needed to examine unique challenges people with lower income may experience. Third, this study was conducted with adults residing in the US, making it difficult to determine whether these results translate to people with CMT living outside of the US. Research is needed to consider the challenges faced in other countries, where there are differences in healthcare practices and policies. Fourth, while the prevalence of CMT does not differ based on racial and ethnic groups,[33] our sample was predominantly White. Therefore, future research is needed to determine whether these findings extend to individuals with other racial or ethnic identities.

5. Conclusion

This study identified and characterized CMT-related challenges that are viewed as important and impactful among individuals with CMT. Results identified nine CMT-related difficulties that are important to individuals with CMT and are often left out of wellbeing assessments in this population. These results can be used to inform future research and clinical practice in the CMT population. As one of the first studies qualitatively examining wellbeing in people with CMT, this study lays the groundwork and provides valuable directions for future research on wellbeing in this population.

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Ethics statement

Our study was approved by Washington University in St. Louis IRB (IRB# 202109166). All participants provided informed consent prior to participation.

CRediT authorship contribution statement

Payton D. Rule: Conceptualization, Methodology, Investigation, Resources, Data curation, Writing – original draft, Visualization, Formal analysis, Writing – review & editing. **Megan W. Wolk**: Formal analysis, Writing – review & editing. **Patrick L. Hill**: Conceptualization, Methodology, Investigation, Resources, Supervision, Project administration, Funding acquisition, Formal analysis, Writing – review & editing.

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence

the work reported in this paper.

Data availability

The data that has been used is confidential.

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Declaration

Recruitment for this study was completed in collaboration with the Charcot-Marie-Tooth Association.

The first author of this manuscript identifies as disabled and holds a volunteer role with the Charcot-Marie-Tooth Association. We have no other conflicts of interest to disclose.

Appendix

Quantitative measures included in the current study survey.

Construct	Measure	# of Items
Sense of purpose	Life Engagement Test[34]	6
Life satisfaction	Satisfaction with Life Scale[35]	5
Health-related quality of life	Official Short Form-36[5]	36
Perceived social support	Multidimensional Scale of Perceived Social Support[36]	12
Self-efficacy	University of Washington Self-Efficacy Scale[37]	6
Discrimination	Everyday Discrimination Scale[38]	9
CMT-related anxiety	Scale created for the current study. Participants were asked to rate how much anxiety they experience about CMT-related things on a scale of 1 (None) to 5 (A lot).	6
CMT/Disability Community Perceived Support	Scale created for the current study. Participants were asked about their sense of connection and belonging to the CMT and broader disability communities.	5
CMT/Disability Community Involvement	Participants were asked if they had been involved with the CMT or broader disability communities within the past two years.	1
Adaptive device use	Participants were asked if they utilize any adaptive devices and, if so, how helpful their adaptive devices are.	2

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