ORIGINAL ARTICLE

Myotonic Dystrophy and Huntington's Disease Care: "We Like to Think We're Making a Difference"

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ABSTRACT: ***Background***: Patient-centered care for individuals with myotonic dystrophy (DM1) and Huntington's disease (HD)—chronic, progressive, and life-limiting neurological conditions—may be challenged by patients' cognitive and behavioral impairments. However, no research has explored health care providers' (HCPs') perspectives about patient-centered care provision for these patients along their disease trajectory. ***Methods***: Constructivist grounded theory informed the iterative data collection and analysis process. Eleven DM1 or HD HCPs participated in semistructured interviews, and three stages of coding were used to analyze their interview transcripts. Codes were collapsed into themes and categories. ***Results***: Three categories including *an evolving care approach, fluid roles*, and *making a difference* were identified. Participants described that their clinical care approach evolved depending on the patient's disease stage and caregivers' degree of involvement. HCPs described that their main goal was to provide hope to patients and caregivers through medical management, crisis prevention, support, and advocacy. Despite the lack of curative treatments, HCPs perceived that patients benefited from ongoing clinical care provided by proactive clinicians. ***Conclusions***: Providing care for individuals with DM1 and HD is a balancing act. HCPs must strike a balance between (1) the frustrations and rewards of patient-centered care provision, (2) addressing symptoms and preventing and managing crises while focusing on patients' and caregivers' quality of life concerns, and (3) advocating for patients while addressing caregivers' needs. This raises important questions: Is patient-centered care possible for patients with cognitive decline? Does chronic neurological care need to evolve to better address patients' and caregivers' complex needs?

RÉSUMÉ: Les soins dans la dystrophie myotonique et la maladie de Huntington : "Nous aimons penser que nous changeons les choses." ***Contexte :*** Les soins axés sur le patient, prodigués aux individus atteints de dystrophie myotonique (DM1) ou de la maladie de Huntington (MH), des maladies neurologiques chroniques, progressives et réduisant l'espérance de vie, peuvent présenter des défis étant donné la présence de déficits cognitifs et comportementaux chez ces patients. Cependant, aucune recherche n'a exploré les perspectives des soignants (PS) concernant la prestation de soins centrée sur le patient tout au long de l'évolution de la maladie chez ces patients. ***Méthodologie :*** La collecte itérative des données et le processus d'analyse ont été réalisés à la lumière de la théorie « constructiviste ». Onze soignants prodiguant des soins à des patients atteints de DM1 ou de MH ont participé à des entrevues semi-structurées et trois étapes de codage ont été utilisées pour analyser les transcriptions des entrevues. Les codes ont été regroupés en thèmes et en catégories. ***Résultats :*** Trois catégories incluant une approche de soins évolutive, des rôles changeants et la contribution à l'amélioration à la qualité de vie des patients ont été identifiées. Les participants ont décrit que leur approche aux soins cliniques avait évolué selon le stade de la maladie du patient et le degré d'implication des soignants. Selon les PS, le principal objectif est de donner de l'espoir aux patients et aux soignants au moyen de la gestion médicale de la maladie, de la prévention de crises, du soutien et de la défense des intérêts des patients. Malgré l'absence de traitement curatif, les soignants considéraient que les patients bénéficiaient des soins cliniques prodigués par des cliniciens proactifs. ***Conclusions :*** Fournir des soins à des individus atteints de DM1 ou de la MP constitue une question d'équilibre. Les soignants doivent faire l'équilibre entre les frustrations et les gratifications qu'impliquent les soins centrés sur le patient, le soulagement des symptômes et la prévention et la gestion des crises tout en portant attention aux préoccupations concernant la qualité de vie des patients et des soignants et à la défense des intérêts des patients. Ceci soulève des questions importantes : les soins centrés sur le patient sont-ils possibles chez des patients présentant un déclin cognitif ? Les soins neurologiques chroniques doivent-ils évoluer afin de mieux répondre aux besoins complexes des patients et des soignants ?

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Patient-centered care for individuals with uncommon neurological disorders may be complicated by variable symptom presentations, limited treatment options, and a lack of evidence-based clinical management models.[1] Shared decision-making is considered the "pinnacle" of patient-centered care,[2] but researchers and clinicians struggle to define what this means in practice,[3] and whether a patient-centered care approach is always feasible, achievable, or desirable.[4] Previous qualitative research found that clinicians caring for patients with progressive neurological disease face numerous challenges as patients approach end of life; in particular, patients' complex and variable disease presentations complicate prognostication, and it is difficult for clinicians to communicate effectively with individuals who have cognitive or speech impairments.[5] However, end of life is only one phase of chronic neurological diseases; therefore, it is necessary to explore management of patients' complex and evolving needs over a long disease trajectory that may span years or decades. Although there are numerous articles that suggest care guidelines for patients with progressive, inherited neurological conditions[6-10]—to our knowledge, clinicians' perspectives about providing care for these patients along their disease trajectory have not been explored. This knowledge gap is significant because we speculate that a patient-centered care approach may be challenging to enact for individuals living with complex, chronic conditions, particularly those presenting with cognitive or behavioral dysfunction. Myotonic dystrophy (DM1) and Huntington's disease (HD)—genetic, chronic, progressive, and life-limiting neurodegenerative conditions—are well-suited to exploring clinicians' perspectives about caring for patients living with protracted physical, behavioral and cognitive impairments.

Exploring care approaches in DM1 and HD may illuminate care delivery for a range of chronic neurological conditions that also impact mobility, cognition, and social function. DM1, the most common adult muscular dystrophy, is a multisystem disorder affecting the muscular, ocular, cardiac, endocrine, gastrointestinal, and central nervous systems. The clinical manifestations of DM1 include muscle weakness, delayed muscle relaxation, arrhythmias, excessive sleepiness, and early-onset cataracts.[11] There is a well-recognized DM1 personality pattern described in the literature, suggesting that patients may have low IQ and apathy levels[12]; in turn, patients may be noncompliant, miss clinic appointments, and seem disinterested in their health.[13,14] Similarly, although HD is characterized by impaired motor function, it is the psychiatric issues—including depression, apathy, anxiety, obsessions and compulsions, impulsivity, irritability and aggression, and psychosis—that are far more debilitating to patients and their families.[7,8] Cognitive impairment variably affects individuals with HD, but when present, results in difficulty with executive function and problems acquiring, processing, and remembering information.[15] Therefore, patients may be unaware of the extent of their symptoms and deficits. Both conditions are life-limiting, and pneumonia is a common cause of death in DM1 and HD.[16-18] Additionally, DM1 patients may experience sudden death secondary to choking or a cardiac event,[16,17] and the suicide rate in HD is higher than the general population.[19]

There are no treatments for DM1 or HD that slow or reverse neurodegeneration, and there is no cure for either condition. However, there are strategies to manage symptoms, and the goal for treating individuals with DM1 and HD is to increase quality of life to "reduce the burden of symptoms, maximize function, and to eliminate unnecessary 'surprises' as affected individuals pass expected disease milestones."[6] Because of the relative rarity of DM1 and HD, most family physicians—and many neurologists for that matter—have little experience managing these patients; thus, care is often provided by specialty neuromuscular or movement disorder clinics.[14] The literature suggests that a multidisciplinary approach that supports the individual and his or her family along the disease trajectory is an "ideal" care plan for individuals with DM1[10,14] and HD.[6,20] Chouinard et al[14] proposed a DM1 management model that considers the multisystem nature of the disorder; the propensity for individuals to have limited educational, economic, and social opportunities; and the lack of knowledge of nonspecialist health care providers (HCPs). Similarly, Nance[6] proposed the "HD molecule" as a model for HD care: the patient and his or her family members are at the center of complex care needs including symptomatic and crisis management, family issues, education, and support that—ideally—should be addressed at each visit by a multidisciplinary care team. Treatment plans may include medications to alleviate symptoms, referrals to specialists to monitor complications, and assessments by physical therapy, occupational therapy, or social work to address activities of daily living and quality of life.[7-9,20] However, treatment approaches may be complicated because symptom presentation, severity, and disease course vary by individual, even among individuals within the same family. Patients' progressive functional decline may further challenge clinical approaches, and family members may therefore become instrumental for monitoring an individual's physical and behavioral changes and for ensuring that he or she is following treatment recommendations.[8]

There are no studies that explore clinicians' approaches to patient-centered care for patients with DM1 or HD along their disease trajectory; therefore, it is unknown if current care approaches are optimizing patient-centeredness and if patients' and caregivers' concerns are being heard and adequately addressed. The challenges of caring for these complex patients warrant investigation because findings may have health care delivery implications—not only for patients with DM1 and HD—but also for patients with other chronic diseases that present with cognitive or behavioral impairments. This study explored how health care professionals perceive of and provide care for individuals with DM1 or HD throughout their disease course. How do HCPs approach the care of these individuals, how effectively do they feel they are meeting patients' needs, and does their approach to care evolve over the course of the illness?

Methods

This analysis is part of a larger study that explored perceptions about clinic attendance for individuals living with DM1 and HD (authors, in preparation). Briefly, the iterative data collection and analysis process were informed by constructivist grounded theory, a qualitative research methodology that studies how basic social processes (e.g. clinical care for a chronic health condition) evolve over time.[21,22] In the larger study, we purposively sampled patients with mild to moderate DM1 or HD (by physician report), their caregivers, and health care professionals at an academic medical center in Ontario, Canada. Table 1 provides a snapshot of

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Table 1: Clinical care for DM1 and HD patients at one academic center in Ontario, Canada

	DM1	HD
Clinical cohort	• Approximately 150 patients are followed every 6 months to 2 years.	• Approximately 125 patients are followed every 3 to 6 months.
Clinical context	• DM1 patients are seen in a general adult muscle disease clinic.	• HD patients may be seen in the monthly HD multidisciplinary clinic or • in a general movement disorders clinic.
Staff/personnel	• All DM1 patients are followed by one neuromuscular specialist. • One nurse practitioner follows most adult neuromuscular patients. • Medical students, residents, or fellows may participate in care.	• Four neurologists specialize in movement disorders; one staffs the multidisciplinary clinic. • A psychiatrist and social worker staff the multidisciplinary clinic; their services are also available by referral. • There is not a nurse affiliated with the HD multidisciplinary clinic. • Medical students, residents, or fellows may participate in care.
Management	• Genetic testing: Confirmatory testing only; referral to genetics for presymptomatic counseling and testing. • Neurological examination, evaluation of cardiac, respiratory and swallowing symptoms, and assessment of psychosocial needs. • Medical treatment: stimulants for fatigue, BiPAP, CPAP • Annual ECG • Referrals may be made to cardiology, respirology, speech language pathology, occupational or physical therapy, or other specialists or allied health professionals. • Patients may be referred for a palliative care consult and/or admitted to a nursing or group home. However, this is relatively rare, because DM1 does not a defined "late" or "end stage."	• Genetic testing: Confirmatory testing only; referral to genetics for presymptomatic counseling and testing. • Neurological examination, evaluation of motor and psychiatric symptoms, and assessment of psychosocial needs. • Medical treatment: antidepressants or antipsychotics to treat psychiatric symptoms; tetrabenazine to manage chorea. • Referrals may be made to speech language pathology, occupational or physical therapy, or for psychiatric or psychological care. Patients may also see the HD social worker or psychiatrist independently from regular neurological follow-up. • At late or end stage of disease, patients may be referred for a palliative care consult or admitted to a nursing home. These patients may be lost to follow-up because they are either unable to come to clinic and/or they are institutionalized.

BIPAP = bilevel positive airway pressure; CPAP = continuous positive airway pressure; ECG = electrocardiogram; HD = Huntington's disease; MD = myotonic dystrophy.

the clinical contexts. Participants were invited to participate in semistructured interviews; 14 patients (n = 5 DM1), 10 caregivers (n = 2 DM1), and 11 HCPs including five neurologists, a psychiatrist, a respirologist, a nurse practitioner, two social workers, and a physiotherapist consented. We ceased recruitment when we determined that the collected data were sufficient to provide a robust exploration of participants' experiences of caring for individuals with HD and DM1. This study reports on the data collected from HCPs; patient and caregiver data will be reported elsewhere.

All interviews were audio-recorded and transcribed verbatim; in turn, each line or sentence of the first two transcripts was coded using words or phrases that captured the experiences or actions described by the participants (initial coding). Next, the most frequently occurring codes were consolidated into preliminary categories and used to determine a focused code for the next six transcripts to determine their fit and relevance. The research team met frequently to discuss preliminary findings, and the final list of categories was developed by consensus and used to recode the entire dataset (theoretical coding). Throughout the research process, data within and between transcripts were constantly compared, and the research team wrote memos and drew diagrams to capture and explicate increasingly abstract ideas about the data. Nvivo, a qualitative research software program, was used to organize and manage the data. The Western University Research Ethics Board approved the study. To protect confidentiality, all participants were given a pseudonym.

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Results

Three thematic categories including *an evolving care approach, fluid roles*, and *making a difference* were identified. The participants described that their approach to care was dependent on the patient's disease stage and the presence of caregivers and their degree of involvement. HCPs also perceived that their role in care evolved over the disease trajectory, encompassing educating patients and families, preventing crises, and providing medical management, support, and advocacy. In the absence of disease-halting or curative treatment, these roles contributed in various ways to an overarching goal of providing *hope*. Reflecting on their efforts to make a meaningful difference to patients and their families, HCPs described the rewards and challenges of their care approach; in particular, although participants expressed frustration and a sense of futility about their inability to provide a cure, they perceived that patients benefited from regular follow-up with proactive clinicians.

An Evolving Care Approach

First Contact

Patients are typically referred to a specialty neurology clinic for three reasons: individuals are at risk for inheriting DM1 or HD; individuals are gene positive but presymptomatic; or individuals have neurological symptoms with or without a family history. Initial visits typically include a review of symptoms and family history and a comprehensive neurological examination. Clinicians perceive that patients come to seek a diagnosis and information from a specialist regarding symptoms, genetic status, or the impact that the disease will have on their families. Family members often attend the initial visit not only to support their family member, but also because there is a—perhaps unspoken—expectation that they will also receive information and counseling. Patients who do not know their gene status or who are gene positive but presymptomatic may be apprehensive about their initial clinic visit:

> I'm kind of the physician that people hate to meet…There was one lady who was a runner, and literally her husband came bolting upstairs…and said 'have you seen my wife?'... He tried to drop her off…and she just bolted…because coming to see me is potentially coming face-to-face with your genetic fate. (Dr. Green, neurologist)

A Flexible Approach to Follow-up

The approach and content of the follow-up visit varies at each time point and continues to evolve once HD and DM1 patients begin to manifest symptoms. A typical visit for both DM1 and HD consists of a neurological examination including a review of symptoms and a functional assessment, followed by a discussion about treatment options and research opportunities. Medications —including stimulants to treat excessive fatigue in DM1, or antidepressants for the psychiatric manifestations of HD—are available to mitigate symptoms, but are generally only prescribed if the patient is experiencing decreased quality of life. Patients and caregivers are then given the opportunity to ask questions or to have their concerns addressed.

> We would talk about any of the cognitive issues, memory problems, any behavioural change, any problems with interpersonal with their working, or at home with the family. And, then we'd review generally how they're doing, functioning, working, how work is going, how are things going with the family, plans and things long-term. We might talk about driving issues, if that were a problem. And, then any other, obviously starting out first with any concerns they have or any issues. Usually a caregiver would come and I would also speak with the caregiver about how things are going. (Dr. Roberts, neurologist)

Although the clinicians' approaches were similar at both clinics, the structure of care and the role of allied health professional participants differed. The neuromuscular clinic team has access to allied health professionals including physical therapy and social work, but these clinicians cover a large spectrum of neurological illness and do not specialize in DM1. DM1 patients are therefore only referred to a physiotherapist on an as-needed basis: "So, I don't follow them. I don't follow their progression. I don't know when they're coming" (Diane, physiotherapist). Similarly, the social worker typically works with DM1 patients on a one-time referral basis to assist with disability paperwork. In contrast, the social worker is an integral member of the multidisciplinary HD clinic. In addition to providing care outside of the clinical setting, the social worker evaluates patients concurrently with a neurologist and a psychiatrist during clinic visits:

> … it's an extremely thorough dialogue and what I really like about it is, it's not your typical medical model. It's much more of a bio-psycho-social … not just the physical function of the person but how they're interrelating with their work, peers, how they're interrelating with their family members, how is it affecting their quality of life… (Ray, social worker)

Although HCPs seemed to have standard templates for how they conducted initial and follow-up visits, they described encouraging patients and their loved ones to direct the focus of the clinic visit and to be actively engaged in making health care decisions. However, HCPs described that this had variable efficacy because patients were sometimes unaware or disinterested in addressing symptoms that could lead to morbidity and mortality; in particular, patients' progressive cognitive and behavioral functional decline directly impacted their ability to recognize and address problematic symptoms. Participants stated that family caregivers became increasingly important for addressing concerns and making decisions:

> … It's often significant, again in the patients that have cognitive involvement because they may have lost the cognitive capacity to understand what's going on, or they've become apathetic so they don't really care. They need somebody to motivate them to do all the right things like take the medications they're prescribed for other conditions, to understand why they need investigation for certain things, and why they need to go to other appointments. It's very helpful to have a caregiver there to help them do all of those things. (Dr. Matthews, neurologist)

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Sometimes, a conversation with patients about the purposes and goals of follow-up visits was perceived as mutually beneficial:

> So, I ask the patient, what do they think the visit is for, do they have any questions, concerns, things that they specifically want to address, they want me to address, and then I'll address those. I generally will do a physical exam and surveillance for their breathing, cardiac, swallowing, speech and how they're managing at home in terms of a functional perspective. That's within the realm of whether or not they're interested in that and sometimes their goals will be, 'I was just told to come here, and so I'm coming.' And, so, then we have to discuss that as well. (Martha, clinic nurse)

Fluid Roles

One participant described that neurologists perceive that they are the "quarterback" (Dr. Matthews) who is responsible for directing all aspects of the patient's care, with support from nurses, specialists, and allied health professionals. Participants recognized the lack of curative treatments for DM1 and HD, and therefore defined their role as primarily one of providing hope by: (1) providing expert evaluation and education; (2) preventing and managing crises, (3) being an advocate, and (4) providing support. These roles were not seen as mutually exclusive; rather, they were perceived as interrelated and evolving over time. In particular, the HCPs perceived that discussing research opportunities, offering symptomatic management options, and reassuring patients about their functionality provided patients with the sense that "… somehow in seeing us, we're dealing with the active disease and, in seeing someone, something's being done to help treat them" (Dr. Roberts, neurologist). Clinicians also sought to ease patients' isolation and to reassure them that the wider medical and research communities had not forgotten them:

> One of the things I see as my job is to let [them] know that no, actually there is a lot of stuff that's happening. I think they're starting to see it now in the sense that 10 years ago [their former doctor] didn't talk to them about clinical trials in Huntington's disease because there weren't any; whereas, now we've got a couple of research opportunities for you if you're interested…. I think they do get some hope from that. (Dr. Green, neurologist)

Providing Expert Evaluation and Education

Before and following diagnosis, participants stated that the primary role of the specialist clinician was to provide education and guidance to patients and family physicians, while providing surveillance for emerging or worsening symptoms. Participants described the importance of specialists' expertise in relation to family physicians' lack of familiarity with these uncommon conditions, and perceived their role as one of educating family physicians and augmenting primary care:

> Some family docs are quite knowledgeable, or take it upon themselves to learn a little bit about the disorder. But when you think that it's really 1 in 8000, not every family physician will have an individual or a family with myotonic dystrophy….it's an uncommon disorder when you think about all the other things that family physicians have to deal with… So, my role… is that our clinic notes serve as a guide or a template for what needs to be watched for. (Dr. Thompson, neurologist)

Patient education largely involved describing inheritance patterns, symptoms, and the variability of disease progression. Providers also supplied patients with pragmatic information regarding management strategies (e.g. breath stacking to improve respiratory function for DM1 patients), assistive devices, and funding or support resources. Some HCPs prioritized keeping abreast of the latest research and pharmaceutical options then distilling information for patients. Although HCPs perceived most patients to be active participants in their care, they identified that information seeking was often a more important priority for caregivers, especially as the patient's condition deteriorated. Therefore, HCPs made judgment calls about the amount, content, and timing of information that was given to patients at different points along the disease trajectory.

> I try to encourage them to ask questions because at the first visit after I say you've got Huntington's disease, I could yammer on for another 20 minutes, but they hear nothing because they're just stuck on 'I've got HD'...that's led me to not give them too much information the first time because it's going to have to be reinforced on subsequent visits … (Dr. Green, neurologist)

Preventing and Managing Crises

In the absence of treatments to reverse or slow disease progression, HCPs stated that their treatment approaches centered around preventing complications and managing crises. Clinic visits were an opportunity to monitor symptom progression and order tests or refer to other specialists to evaluate potentially life-limiting complications.

> The reason that I've gotten into the care of that patient population [DM1] here is because … We want to identify patients that potentially need some type of breathing support for the rest of their life and can we identify that group that's going to do well … and then try to look for those resources. (Dr. Vincent, specialist physician)

The HCPs described that an essential part of their role was to be flexible about care approaches and to put structures in place to enable a rapid response to serious and acute issues. Specialists and allied health professionals were aware that mobility and transportation difficulties complicated patients' ability to come to a clinic, and therefore made efforts to accommodate patients. Strategies included evaluating the patient in tandem with other doctor's appointments, making house calls (HD social worker), having nurses or social workers respond quickly by phone to

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emergencies or acute issues, and to 'squeeze' patients into clinic for acute needs.

> …if they're coming to, say, see the doctor, and we know they need this done, there might be transportation issues or distances, so then I'll offer them up I can do it on a day they're coming for other tests or other doctors' visits if they could wait that long. But, if it's a whole year, I try and get them in just on my own day or see them when they're coming to see Dr. Vincent or see them when they're coming to see Dr. Thompson or Martha. (Diane, physiotherapist)

Being an Advocate

Participants emphasized the importance of advocating for patients by raising community awareness about these uncommon conditions. HCPs described "being a spokesperson" for DM1 or HD by participating in charity events and giving talks at support groups or patient education conferences. Clinicians—particularly nurses and social workers—were instrumental in helping patients obtain funding and community resources. "Martha (*nurse*) is very good at connecting people…I think Martha is the linchpin…for linking people to resources" (Dr. Thompson, neurologist). HCPs sometimes acted as a liaison between the patient and his or her employer, family physician, or family member. In particular, clinicians tried to balance caregivers' concerns with being a 'voice' for the patient and encouraging the patient to express his or her needs.

Providing Support

Similarly, supportive care meant a number of things to participants including providing counseling to help patients and families adjust to the diagnosis and strategies for managing the disease as it progressed, seeking resources for patients and their families, or offering guidance for family physicians to provide primary care for HD or DM1 individuals. Specialist physicians perceived an inverse relationship between their role and the patients' disease progression; that is, as patients begin to deteriorate and options for symptomatic treatments diminished, allied health professionals become increasingly important for obtaining resources and to helping patients and their families cope with behavioral and cognitive changes.

> Dr. Green will often say that…I'm more important to be at these clinics than he is because it's [HD] much more of a psychosocial disease and there's not a whole lot that can be done likely to stop the disease at this point. He can manage some of the symptoms but it's important for someone, like myself, to be around to help manage all the social challenges that come along with the disease. (Ray, social worker)

Making a Difference

Frustrations

The variable and unpredictable features of DM1 and HD—namely the behavioral and cognitive impairments—frustrated practitioners and challenged their ability to prognosticate and to provide education and care. These frustrations were exacerbated by a lack of resources including limited funding and community resources, few treatments and research advances, and lack of time to address patients' complex care needs. Clinic time was limited and some HCPs had a backlog of patients requiring initial consultations and follow-up; consequently, providers were not always able to address patients' multiple physical and psychosocial needs. Moreover, there was limited funding available for allied health professionals to provide supportive services:

> There's probably a greater need for these services… particularly social work, speech and swallowing. … Certainly the social work position that we have funded…is only a part-time position. I'm quite suspicious that he does more than 1 and a half days per week, but that's all he gets paid for. (Dr. Green, neurologist)

HCPs devoted a significant amount of their limited clinic time providing education about DM1 and HD, including management strategies to mitigate symptoms. HCPs described that patients did not retain information, and some were either unaware—or apathetic about—the importance of following treatment recommendations:

> They are draining in the sense that, you can see them year after year after year and nothing has changed, they're still eating like they're not supposed to, they're still smoking and they're not supposed to, they may or may not take care of themselves, and that's just the way they are. So, I find them a significant challenge to take care of. (Martha, clinic nurse)

HCPs expressed a sense of futility that despite their best efforts, patients' function, and quality of life would continue to deteriorate:

> We do contribute for sure because we manage fairly complicated aspects of the disease, including, for example, the behavioural aspects… It is not satisfying in any way because this is a progressive, relentless degenerative disease and we can't do anything. Unlike, for example, dystonia, torticollis, blepharospasm, or even Parkinson's disease, where we have excellent medications that can improve the quality of life for potentially 20 years. (Dr. Bennett, neurologist)

Rewards

These challenges were tempered by the perceived rewards of caring for individuals with DM1 or HD. Most HCPs stated that they pursued a career in health care because of an intrinsic desire to make a difference in peoples' lives. Moreover, the specialist physicians were inspired—and rewarded—by the intellectual challenge of diagnosing and managing complex neurological disease. Therefore, despite a sense of futility, HCPs received small and intangible benefits by being proactive about providing care for patients and families. In essence, HCPs believed that providing support and advocacy was the "right thing to do" in the absence of other therapeutic options:

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> Neurology seems to be a specialty that you could make a difference in, despite not having curative treatments for a lot of the conditions that we have. So, having a relationship with patients and families was an important aspect of neurology as a career path. (Dr. Thompson, neurologist)

One participant described that caring for these patients filled a void in care that had not previously been addressed by his departmental colleagues: "I didn't think that our [specialist] group was providing them with all the necessary service that was required to properly care for this population [DM1]" (Dr. Vincent, specialist physician). Other participants enjoyed forming long-term relationships with patients, having the "privilege" to care for multiple generations of a family (Dr. Green, neurologist), and guiding patients through difficult and emotional situations:

> … you follow these people along for years you get to know them. I've followed several mothers through pregnancies. Another whole issue is the genetic counselling of a woman in childbearing years about the risks that she might have an affected child ….There is some reward in being able to take somebody through that, even though you can't actually treat the disease. (Dr. Matthews, neurologist)

Discussion

> We do not know whether or not it makes any difference to their life, quality of life, or their health, if they come to the clinic or don't come to the clinic… We like to think we're making a difference, but I don't think we know that. (Martha, clinic nurse)

The lack of literature exploring clinicians' perspectives about patient-centered care provision for patients with DM1 or HD left us with similar questions: how do health care professionals describe their management approach for these patients, and do they believe that the current patient-centered clinical care models meet patients' needs? Participants described the necessity and importance of providing proactive, expert, evolving, and ongoing care in specialty neurology clinics because they recognized that the uncommon prevalence of DM1 and HD and limited system resources precluded primary care HCPs and generalists from addressing patients' and caregivers' complex needs. However, similar issues—coupled with patients' variable disease presentation—also challenged specialist HCPs' abilities to provide patient-centered care. Researchers seeking to develop a chronic care model for neurological conditions interviewed 180 HCPs, community members, and policy makers, and identified similar challenges.[1] Although this study comments on general care for a number of neurological conditions, it does not specifically explore the perspectives of providing patient-centered care for DM1- and HD-affected individuals along their disease trajectory. Our findings suggest that a patient-centered care provision for patients with DM1 and HD is a balancing act that evolves over the course of the illness; in particular, HCPs must consider whether system capabilities, including time, funding, and their medical training, afford them the opportunity to address concerns that are most important to patients and their families.

HCPs must constantly temper their frustrations and sense of futility with the perceived rewards of caring for individuals with DM1 and HD. Although our participants had expert knowledge about these conditions and were able to form long-term relationships with patients, they described that they were frustrated by the lack of pharmaceutical treatments, limited community resources and funding opportunities, overburdened clinics with long wait lists, and the emotional cost of caring for these patients. Participants recognized that the emotional, physical, and social implications of DM1 or HD diagnosis and symptomatic progression impact the entire family,[23-27] and that emotional support may be one factor that buffers the impact of caregiving stress.[28] But research participants described that they had limited time to address caregivers' needs, which resonates with the experiences of other providers caring for patients with chronic neurological conditions.[1,29] This raises questions about whether it is necessary —or feasible—for a specialty neurology clinic to devote resources to provide ongoing counseling and support to nonaffected family members.[23,24,26] Although these resources are available in some clinical contexts,[30,31] they are not universal,[28] and the current physician-led model at our academic center is not designed to provide this degree of social support. In the current fee-for-service model in Ontario, specialist physicians are paid for services provided only to the individual referred for consultation, therefore—although our participants described that they spent considerable time addressing caregivers' needs—there are limited opportunities for remuneration for this important work. Similar challenges —particularly time-limited appointments and a lack of adequate resources to support allied health professionals—were echoed in a small qualitative study examining the perspectives of physicians caring for patients with Alzheimer's disease.[32] In essence, there was a sense amongst our participants that the current physician-led care model at our academic center was not doing "enough"; that is, participants were unsure if their care approach was making a difference in patients' and caregivers' daily lives. As Dr. Thompson (neurologist) stated, "we can do a lot better."

HCPs also seemed to struggle striking a balance between their role as a patient advocate and their reliance on, and need to support, caregivers. Although participants stated that they encouraged patients to direct the clinical encounter, they raised concerns that patients' progressive cognitive decline and behavioral impairments challenged education and symptomatic management, and they had to rely on caregivers' to provide health information as patients' health deteriorated. Moreover, the hereditary nature of DM1 and HD—and the complex care needs patients require as they progressively decline—requires health care professionals to address the needs of caregivers and those at-risk.[8] In addition, our findings resonate with previous literature[33,34] that suggests that issues of clinical concern to HCPs may not be what patients and caregivers are aware of or want to address. Although HCPs focus on symptoms that may cause morbidity or mortality, patients and caregivers are generally concerned with issues that impact their relationships[26] and participation in education, employment, and leisure and recreational activities.[35] We speculate that our physician participants perceived that they were qualified to treat symptoms, but were less comfortable addressing patients' and caregivers' social and quality of life issues. This has important implications for treatment approaches and raises questions about

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patient-centered clinical care models: Is care still "patient-centered" if HCPs address issues they know to be important, even if they are not prioritized by patients and families? Are HCPs trained adequately to maintain patient-centered care as patients functionally decline? Finally, are health care teams sufficiently nimble to engage allied health professions who may be better equipped to address patients' evolving needs?

Reconciling these questions and complications to create a clinical model that is responsive to patients' and caregivers' needs is challenging. The multidisciplinary team at our institution's HD clinic seemed to alleviate some of these challenges. In contrast, the neuromuscular specialists described that there was not a structure in place to support a multidisciplinary DM1 clinic, and they differed in their beliefs about the feasibility and utility of creating one. Further, our participants suggested that—in the absence of disease-halting or curative treatments—that their main role was to provide hope for patients at each stage of their disease process through education, advocacy, support, and medical management.

We question, however, whether the traditional "neurologist-as-quarterback" clinical model described by our participants is the most efficient and effective model for providing hope and for addressing the evolving needs of patients and caregivers. In fact, some participants suggested that the physician's role may become less useful as patients deteriorate because the issues that physicians are best trained to address may not be the issues that concern patients and caregivers most. Too often, HCPs efforts to make a difference in patients' and caregivers' lives may focus on the "margins" of the illness experience—those things that are readily addressed by traditional medical models of care, such as providing education about illness, prescribing medications to treat symptoms, and assessing and preventing complications. We are not suggesting that these efforts are not important. Rather, we are suggesting that these efforts may be insufficient, and may miss critical opportunities to make a difference in patients' and caregivers' daily lives.

Participants in the present study recognized that nurses and allied health professionals become increasingly important as patients' symptoms progress, perhaps because their expertise is better aligned to the needs of patients and families with evolving chronic illnesses. We speculate that alternate models of care and leadership, including family health care teams, or rehabilitation or nurse-led multidisciplinary clinics, may have merit in these populations. Research suggests that a chronic care model for neurological conditions should be an "intersectoral collaboration" between policy makers, community members, and the health care system.[1] Multidisciplinary, nurse-led clinics are the standard model for other chronic disease populations including heart failure,[36] cancer,[37] and diabetes,[38] and research suggests that patients attending nurse-led clinics have improved self-care behaviors and/or better outcomes. Although physicians remain integral to diagnosis and treatment, nurses may be ideally suited to providing holistic, patient-centered care for patients and their families along the disease trajectory; that is, nurses are able to monitor symptoms and treat complications while also providing education, advocacy, and ongoing support. Nurses are trained to see—and provide emotional support—to the patient and caregiver as a unit, and may therefore be best-suited to creating a comfortable clinic space where patients and caregivers can have their complex biopsychosocial needs addressed.

A nurse-led model for the care of patients with DM1 has been proposed,[14] and work is currently being done to create and assess a nurse-led, integrated clinical care pathway for DM1 at a neuromuscular clinic in Quebec. The DM1 clinic is led by a nurse care manager who works with an interdisciplinary team to fulfill the essential care roles identified by our study participants; that is, the nurse care manager monitors symptoms, treats complications, educates, and supports the psychosocial needs of the patient and his or her family.[39] Several clinics for DM1 and HD in the United States are funded by patient advocacy groups that support a multidisciplinary group of clinicians to provide care and to present research opportunities to patients and families.[30,31] However, to our knowledge, although multidisciplinary, nurse-led or the chronic care clinical models show promise, they have not yet been systematically evaluated or implemented across North America.[1,39]

Limitations and Future Directions

This is a small study describing the perceptions and experiences of HCPs caring for patients with DM1 and HD at one Canadian academic center. We recognize that clinics and treatment approaches may vary at different locations, and our highly contextualized study is therefore not generalizable to other settings. Additionally, study participants described the challenges of supporting family physicians to care for DM1 and HD in the community. We did not, however, interview any family physicians about their perceptions of, and approaches to, providing primary care for these individuals. Similarly, we suggest that nurse-led multidisciplinary clinics might be a useful model of care for DM1 and HD patients, yet recruitment challenges and the limited number of nurses specializing in DM1 and HD at our institution precluded greater nurse participation. Because care for patients with DM1 and HD is complex, future research should explore the perspectives of nurses, family physicians, and other allied health professionals. Given our findings that HCPs have limited time to address the social and emotional impact of DM1 and HD, future research should explore the utility of adding a psychologist with expertise in chronic, inherited conditions to the multidisciplinary care team. Finally, it is essential to understand the experiences and health care expectations of DM1 and HD individuals and their families before proposing a model of care.

Conclusion

Despite challenges providing patient-centered care, HCP participants perceive that DM1 and HD patients benefit from clinical follow-up with expert clinicians who are proactive about managing complications, providing support, and conveying hope. However, our findings suggest that patients' and caregivers' needs may not be sufficiently addressed by traditional physician-led clinical models. Participants identified a need for greater involvement from allied health professionals, and we suggest that nurses are integral for enacting a holistic care approach. It may also be necessary to modify current medical education curricula and resident training programs to ensure that clinicians are better equipped to holistically integrate the complex needs of patients living with chronic disease into collaborative practices. Nurse educators and experienced nurse clinicians may be well-suited to address this critical training gap. Regardless, we propose that research exploring clinical models for patients with complex physical, cognitive,

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and behavioral needs is warranted. We anticipate that our findings will add to scholarly conversations about patient-centered care for patients with complex chronic conditions, and that our findings may resonate with, and inform, care practices for various patient populations who experience unrelenting, chronic, and progressive physical and cognitive decline.

Statement of Authorship

KAL designed the study, collected and analyzed the data, and wrote the initial and subsequent drafts of the manuscript. CJW contributed to data analysis and manuscript preparation. SLR contributed to study design and data analysis. CP contributed to data collection and manuscript preparation. SL Venance contributed to study design, data analysis, and manuscript preparation.

Disclosures

The authors have no conflicts of interest to disclose.

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