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Are Patients at the Centre of Care?: A Qualitative Exploration of Myotonic Dystrophy Type 1 (DM1)

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A thesis submitted in partial fulfillment of the requirements for the degree in Doctor of Philosophy

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ARE PATIENTS AT THE CENTRE OF CARE?: A QUALITATIVE EXPLORATION
OF MYOTONIC DYSTROPHY TYPE 1 (DM1)

(Thesis format: Integrated Article)

by

Kori A. LaDonna

Graduate Program in Health & Rehabilitation Sciences

A thesis submitted in partial fulfillment
of the requirements for the degree of
Doctor of Philosophy

The School of Graduate and Postdoctoral Studies
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Abstract

Health care for individuals living with myotonic dystrophy (DM1)—an uncommon, life-limiting neurological condition for which there are few treatments—may be challenged by patients’ symptoms including cognitive and behavioral impairments. Is patient-centered care—which incorporates the values, experiences and expertise of patients and their caregivers—feasible or achievable? Uncovering patients’ and their caregivers’ experiences of living with DM1, their health care expectations, and their health care providers’ (HCP) perspectives about care is essential for examining patient-centered care in this population. Therefore, the purpose of this research is to: (1) add patients’ and caregivers’ voices to the literature, (2) explore on-going care provision for individuals with DM1, and (3) probe whether patients’ and caregivers’ needs are being met.

Forty-nine participants were purposively sampled from one academic centre in Ontario, Canada to participate in three studies. Phenomenology, photovoice and grounded theory—qualitative methodologies that prioritize participants’ experiences and recognize that researchers and participants co-construct the data—were used to explore patients’, caregivers’ and HCPs’ experiences about living— or caring for individuals—with DM1. Semi-structured interviews were the primary data collection method; focus groups and photographs were also used in the photovoice study. Data analysis varied by methodology.

Patient and caregiver participants’ described that DM1 symptoms—particularly fatigue and weakness— impacted their daily activities and sense of self; however, participants were resilient and problem-solved coping strategies. Patient and caregiver participants’ motivations for clinic attendance evolved along the disease trajectory, but most participants perceived that clinic attendance had tangible benefits. HCPs described that their main role was to provide hope for patients and their families. Most importantly, this research revealed that patient, caregiver and HCP participants described clinic as a ‘safe place’ for patients and caregivers to be understood, and to be empowered to take a proactive role in health care.

DM1 participants derived a therapeutic benefit from attending clinic despite providers’ concerns that patient-centered care was challenged by complex biopsychosocial issues. This

research raises questions about whether a physician-led model is the most efficient mode of care provision, or whether other models warrant investigation.

Keywords

Myotonic Dystrophy; caregiver; dysphagia; Huntington's disease; patient-centered care; qualitative research

Co-Authorship Statement

The study design, data collection and primary analysis for this thesis are the work of Kori A. LaDonna. The advisory committee—Dr. Shannon L. Venance, Dr. Andrew M. Johnson, Dr. Susan L. Ray and Dr. Christopher J. Watling—met regularly to provide guidance to K.A. LaDonna, and as noted below, contributed to review and edits of manuscripts representing Chapters III-VII.

Chapter III: All authors advised K.A. LaDonna about study design and methods for data collection and analysis. K.A. LaDonna conducted all interviews and K.A. LaDonna and W.J. Koopman conducted the data analysis concurrently; S.L. Ray and S.L. Venance reviewed the findings. K.A. LaDonna wrote the initial and subsequent manuscript drafts with input and final approval by all authors.

Chapter IV: K.A. LaDonna designed the study. K.A. LaDonna was responsible for data collection; data analysis was completed by both authors. K.A. LaDonna wrote the initial and subsequent manuscript drafts, reviewed and approved by S.L. Venance.

Chapter V: This paper is an analysis of one theme identified in Chapter IV. A. Ghavanini conducted a retrospective clinical chart review. S.L. Venance and A. Ghavanini reviewed participants' responses in tandem with their charts, and K.A. LaDonna consolidated codes into categories and prepared the initial and subsequent drafts of the manuscript. All authors reviewed and approved the final manuscript.

Chapter VI: Study design, data collection and manuscript preparation were the primary responsibility of K.A. LaDonna. C. Piechowicz participated in data collection. S.L. Venance and S.L. Ray assisted with data analysis. K.A. LaDonna wrote the initial and subsequent drafts of the manuscript with S.L. Venance and C.J. Watling contributing to revisions. KAL, SLV and CW reviewed and approved the final manuscript.

Chapter VII: Study design, data collection and manuscript preparation were the primary responsibility of K.A. LaDonna. C. Piechowicz participated in data collection. S.L. Venance and S.L. Ray assisted with data analysis. K.A. LaDonna wrote the initial and subsequent

drafts of the manuscript with S.L. Venance and C.J. Watling contributing to revisions. KAL, CP, SLV and CW reviewed and approved the final manuscript.

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Chapter 1

1 Introduction

“Helping patients to be more active in consultations changes centuries of physician-dominated dialogues to those that engage patients as active participants” (Epstein & Street, 2011, p. 100). ‘Patient-centered care’ is widely accepted as a key component of quality health care (Institute of Medicine, 2014), and health care professionals (HCPs) are taught and encouraged to put patients at the centre of clinical care. However, patient-centered care is poorly defined and variably enacted in practice (Berwick, 2009). What, then, is ‘patient-centered care’, and is it achievable for all patients, particularly those challenged by cognitive and behavioral impairments? To explore this, I used a variety of qualitative research methodologies and methods to explore on-going clinical care provision for individuals living with myotonic dystrophy (DM1). DM1—the most common adult muscular dystrophy—is a chronic, progressive and life-limiting, multi-system neurological disorder in which affected individuals may experience apathy and borderline IQ (Harper, 2001).

Research suggests that care for individuals with muscular dystrophy may be insufficient (Hill & Phillips, 2006); in particular, care for individuals with DM1 may be complicated by patients’ variable symptoms, their complex biopsychosocial needs, few treatment options, or patients’ and health care providers’ divergent goals (Heatwole, 2012; Gagnon et al, 2010; Meola & Sansone, 2007; Udd & Krahe, 2012). Despite these challenges, only two studies have used qualitative research methods to explore perspectives about on-going clinical care for patients with muscular dystrophy (Hartley, Goodwin & Goldbart, 2011; Nätterlund & Ahlström, 1999). Only 18 individuals with DM1 participated in this research, suggesting that DM1 patients’ voices are relatively absent from the literature when compared to patient populations with other uncommon neurological conditions (LaDonna, 2011). There is also insufficient research regarding caregivers’ and HCPs’ perspectives about care for individuals with DM1.

No exploration or evaluation of patient-centred care can be complete without including and accounting for the voices and the experiences of the patients, caregivers and health care providers who are at the centre of that care. The goal of this research is to add DM1 patients', caregivers' and HCPs' perspectives to the literature, and to explore the current clinical care model at one academic centre in Ontario, Canada. Three qualitative research methodologies—phenomenology (van Manen, 1990), photovoice (Wang & Burris, 1994; 1997) and grounded theory (Charmaz, 2006)—that recognize, prioritize and emphasize individual experience were used. In this introduction, I will locate myself within the constructivist qualitative research paradigm, describe how I came to my interest in patient-centered care, introduce DM1 and care provision for these individuals, describe 'patient-centered care', and state my research questions.

1.1 Background

Hereditary neurological conditions like DM1 may have a physical, emotional, and social impact on individuals and their families, particularly when there are few effective treatments and no cure. I speculate that the lack of curative treatments—coupled with DM1 patients' complex and variable symptom presentation—may complicate on-going, patient-centered care provision. However, patients', caregivers', and HCPs' perspectives about patient-centered care provision (and on-going follow-up) in DM1 have not yet been explored. I contend that any exploration is challenged by (1) the relative absence of DM1 patients' voices from the literature, and (2) the lack of a uniform definition or conceptualization of what 'patient-centered care' is, or how it is experienced in practice (Berwick 2009).

1.1.1 Locating the Researcher

My interest in patient-centered care stems from experiences working with patients with neurodegenerative conditions, their caregivers and health care providers in a variety of professional contexts. I began working in the Department of Neurology at the University of Rochester in Rochester, New York as a fifteen-year-old summer student hired to do filing and data entry for research that explored experimental therapeutics for patients with Huntington's and Parkinson's disease. This experience gave me a behind-the-scenes look

at—and an appreciation for—the long, and sometimes arduous, process of clinical trials research. I had the opportunity to work with international leaders in Parkinson's and Huntington's disease research and witnessed first-hand their passion to try to find answers—if not tangible treatments—that would improve the daily lives of their patients.

Their passion sparked my own interest in health research, and I pursued a dual Bachelors of Arts degree in English and Communication with a specialization in Health Communication. Throughout my studies, I continued with secretarial and administrative tasks in a neurology outpatient clinic where I had the opportunity to interact with patients and observe their relationships with their providers. My growing interest in patient-centered health care led to an internship with the Huntington's Disease Society of America (HDSA) where my duties included charitable fundraising, organizing patient education and advocacy events, and serving on the Board of Directors for the HDSA Upstate New York Chapter. In addition, I acted as a support group facilitator for patients and families with Huntington's disease and young onset Parkinson's disease, an experience that illuminated how these conditions impact patients' and families', and underscored their resiliency while living with a chronic and progressive health condition.

My research career began as a clinical research coordinator for studies exploring depression and Parkinson's disease and the natural histories of rare neuromuscular disorders. My work with Dr. Shannon Venance and Wilma J. Koopman, Nurse Practitioner on a groundbreaking project exploring the transition of young men with Duchenne Muscular Dystrophy from the pediatric to the adult neuromuscular clinic introduced me to the impact that qualitative research methodologies and methods may have on exploring illness experiences and health care. This research led me to pursue graduate studies using qualitative methodologies. My doctoral research program has provided insight into the day-to-day experiences of individuals living with chronic neurological conditions, and emphasizes the importance of qualitative research for adding individuals' voices to the literature.

1.1.2 Locating the Researcher Within the Qualitative Research Paradigm

Guba and Lincoln (1994) define a research paradigm as a worldview that guides a researcher's epistemological and ontological approach to inquiry. In particular, epistemology is a theory of knowledge that considers what can be known and the relationship between the 'knower' and the 'known', while ontology refers to the 'form and nature of reality' (Guba & Lincoln, 1994). Common research paradigms include positivism, postpositivism, post-modernism and constructivism. Given my personal and professional experience working with individuals with chronic neurological illnesses, I come to this research with assumptions, biases, and theoretical presuppositions. In particular, I believe that individuals have the best insight into how their disease impacts their lives, and are thus able to provide important information to health care professionals that may improve health care. Further, researchers do not come into the research field without leaving an imprint of their knowledge and interpretations. The interplay between researchers and research participants is not only unavoidable, but is essential for developing authentic and important data.

Therefore, I align myself within the constructivist paradigm. The constructivist paradigm includes a relativist ontological position and transactional/subjectivist epistemological position (Guba & Lincoln, 1994). Ontologically, a constructivist researcher believes that there are multiple realities that change over time. Epistemologically, constructivists believe that data is co-constructed between the researcher and the participant. Findings therefore are subjective, and there is not one objective truth that can be discovered. As a relativist, I acknowledge that meanings are fluid and constructed by the language participants use to describe them (Finlay & Ballinger, 2006).

1.2 Myotonic Dystrophy (DM1)

Myotonic dystrophy (DM1) is a chronic, progressive, inherited and life-limiting neurodegenerative disorder. DM1 is a multi-system condition presenting with central, muscular, endocrine, cardiac, ocular and gastrointestinal system abnormalities; in turn, patients may experience cognitive and behavioral manifestations, muscle weakness,

diabetes, arrhythmias, early-onset cataracts and incontinence (Harper, 2001). Severe disability generally occurs in the fifth and sixth decades of life (Schara & Schoser, 2006), and respiratory failure, cardiac rhythm disturbances, and pneumonia are common causes of death for individuals with DM1 (de Die-Smulders et al., 1998; Mathieu et al., 1999).

DM1 is caused by a CTG repeat expansion in the 3' region of DMPK on chromosome 19q13, and is autosomal dominantly inherited; consequently, each child of an affected parent has a 50% chance of inheriting the condition. Moreover, DM1 is characterized by anticipation and symptoms are highly variable; that is, the condition tends to present earlier and with greater symptom severity in subsequent generations. For example, individuals with shorter CTG repeat expansions may experience mild or minimally impactful symptoms while those with longer expansions may have significant physical disabilities and cognitive impairments (Harper, 2001). Consequently, it is common for entire families to be affected; that is, a mildly affected parent may provide care for a severely affected child, or a non-affected caregiver may help manage the health of multiple family members.

Researchers have developed a DM1 cognitive and personality profile suggesting that patients may have difficulty with executive function (Sistiaga et al, 2010) and display avoidant personality traits (Delaporte, 1998; Meola et al, 2003). However, research examining cognitive and behavioral impairment in adult-onset DM1 is inconclusive. Some studies have found minimal or no difference in intelligence and cognitive function between individuals with DM1 and healthy controls (Gaul et al., 2006; Rubinsztein, Rubinsztein, McKenna, Goodburn, & Holland, 1997; Van Spaendonck et al., 1995). Others, however, have found significant differences (Perini et al., 1989; Perini et al., 1999), and suggest that affected individuals have structural brain abnormalities including white matter lesions (Minnerop et al., 2011; Tanaka, Arai, Harada, Hozumi, & Hirata, 2012) or frontal lobe impairments (Meola et al., 2003; Modoni et al., 2008; Sansone et al., 2007; Tanaka et al., 2012). Cognition and personality may be related to patients' CTG gene repeat length (Perini et al, Sistiaga et al, 2010) suggesting that patients with more severe disease experience greater cognitive and behavioural challenges. However, this was not found in a study conducted with a different cohort (Winblad, Lindberg &

Hansen, 2005). Individuals with late onset disease and mild symptoms (i.e., smaller CTG repeat expansions) may experience progressive cognitive decline (Modoni et al., 2004; Modoni et al., 2008; Sansone et al., 2007) or develop focal dementia (Modoni et al., 2004).

The DM1 cognitive and personality profile suggests that affected individuals have paranoid or avoidant personality traits that render it difficult for them to participate in activities or develop relationships (Delaporte, 1998; Sistiaga et al., 2010; Winblad, Lindberg, & Hansen, 2005). Affected individuals may also experience impaired executive dysfunction related to cognitive inflexibility (Sistiaga et al., 2010), reasoning, planning, attention and verbal and visual memory (Antonini et al., 2006; Modoni et al., 2008; Sistiaga et al., 2010; Zalonis et al., 2010). Depression and anxiety are variably experienced, but may be related to central nervous system involvement (Antonini et al., 2006), or to social withdrawal or maladjustment to living with a chronic and progressive illness (Meola et al., 2003; Minnerop et al., 2011). Causality, however, is difficult to determine. Cognitive dysfunction and personality traits may be an organic part of DM1 (Perini et al., 1999), or the result of limited social or educational opportunities (Bird, Follett, & Griep, 1983; Modoni et al., 2008; Sistiaga et al., 2010).

Regardless, the impact of symptoms on DM1 patients' health related quality of life (HRQOL) have been described (Antonini et al, 2006; Peric et al, 2010, Laberge et al, 2013). Twenty patients with DM1 had significantly lower scores on the short-form 36 (SF-36)—a HRQOL questionnaire that assesses 8 health domains including physical and emotional health (Ware & Sherbourne, 1992)—than the general population (Antonini et al, 2006). In particular, findings suggest that age and disease severity are inversely related to SF-36 scores (Antonini et al, 2006; Peric et al, 2010), and that cognition, fatigue and mood may decrease HRQOL (Laberge et al 2013). Research suggests that individuals with DM1 living in Quebec, Canada had low educational attainment, high rates of unemployment, low income and were highly reliant on social assistance (Laberge et al, 2007). Moreover, DM1-affected individuals may experience disrupted social participation resulting from their difficulty or inability to engage in employment and recreational activities (Gagnon, Mathieu & Noreau, 2007); the social participation of 200

DM1-affected individuals was impacted by factors including problems navigating the physical environment, poor access to government, health or community services, and limited support from family or friends (Gagnon et al, 2008). Additionally, results from a study exploring the lifestyle risk factors for 200 DM1 individuals found that being overweight or obese, using addictive substances and being physically inactive impacted their health. Finally, the clinical manifestations of DM1 including fatigue, weakness, and low educational attainment and socioeconomic status may challenge patients' ability to engage in health promotion behaviors (Gagnon et al, 2013). Symptoms causing decreased HRQOL—for example, constipation, cataracts, and depression— may be amenable to treatment (Peric et al., 2013), but the complex and variable symptom presentations may challenge care for individuals with DM1 (Heatwole et al 2012; Meola & Sansone, 2007). It is therefore imperative to explore DM1-affected individuals' understanding of their health, and the perspectives of caregivers and HCPs providing care for this population.

DM1 families may experience significant psychosocial problems that may not be adequately addressed by HCPs (Cup et al, 2011). Instead, clinicians may preferentially monitor patients' breathing, cardiac, and swallowing symptoms because they are the most likely to cause serious complications or sudden and/or early death. However, these may be challenging to identify and manage due to the multitude of physical, cognitive, and behavioral impairments that patients may experience (Gagnon et al, 2010; LaDonna, Koopman, & Venance, 2011).

There are no treatments to halt the progressive muscle weakness associated with DM1, and health care providers provide surveillance for both symptomatic and asymptomatic complications (Gagnon et al, 2010; Turner & Hilton-Jones, 2008). DM1 is relatively uncommon, affecting approximately 1 in 8,000 individuals (Harper, 2001), therefore few family or generalist physicians follow these patients. However, DM1 is more prevalent in certain areas of Canada, and the prevalence of DM1 in the Saguenay-Lac-Saint-Jean region of Quebec is 30-60 times higher than the worldwide prevalence (Mathieu, De Braekeleer & Prevost, 1990). Consequently, extensive biopsychosocial and clinical management research has been conducted in this region. For example, researchers in

Quebec have proposed that DM1 patients' complex needs—including the potential for individuals to have limited educational, economic and social opportunities—often requires specialist clinicians to follow a systematic surveillance and treatment plan (Chouinard et al., 2009; Gagnon et al., 2007; Gagnon et al., 2010). Researchers have proposed DM1 management and health supervision models that address routine symptomatic surveillance in addition to potential life-threatening complications. At the neuromuscular clinic where this research was conducted, cardiac conduction abnormalities would typically be monitored with an annual electrocardiogram (ECG), and a referral to a cardiologist would be initiated if there were any symptoms or changes on the ECG. Similarly, pulmonary function tests and inquiry about symptoms related to breathing help to anticipate the need for a referral to a respirologist to follow individuals with chronic respiratory failure. Those with excessive daytime sleepiness may be referred for a sleep study to consider the role of obstructive sleep apnea and are often prescribed a central nervous system stimulant (e.g. modafinil or methylphenidate). Individuals with chronic respiratory failure or sleep apnea may benefit from noninvasive ventilation (Turner & Hilton-Jones, 2008). Often, individuals either complaining of, or suspected of having dysphagia are sent for a swallowing evaluation and instructed on strategies for safe swallowing. Cataracts are treated surgically. Health care providers may assess patients' mobility, social and recreational opportunities, employment and financial status, and overall quality of life (Chouinard et al, 2009). Further, individuals with DM1 may require referrals for genetic counseling, community services and individual or family support resources (Gagnon et al, 2007; Gagnon et al, 2010).

However, the published literature suggests that clinical care for these individuals may be complicated by their cognitive and behavioral challenges. There are reports in the literature documenting that DM1 individuals may miss clinic appointments, poorly adhere to rehabilitation treatment recommendations, and seem indifferent about their health (Chouinard et al., 2009; Meola & Sansone, 2007); however, this has not been the experience of others where DM1 individuals are found to be diligent about keeping their appointments (personal communication, Dr. Shannon Venance). To date, there has been no research that explores DM1-affected individuals' motivations for maintaining on-going clinical follow-up.

1.3 Coming to the Question

‘Patient-centered care’ is loosely defined as “providing care that is respectful of and responsive to individual patient preferences, needs, and values, and ensuring that patient values guide all clinical decisions” (Institute of Medicine, IOM, 2014). Patient-centered care approaches are taught in medical school as a marker of good patient-physician communication that may lead to better patient self-management and improved health outcomes (de Haes, 2006). However, de Haes (2006) contends that these improved outcomes may be difficult to attain in practice and that “a more nuanced standpoint has to be taken” (p. 291). It may also be difficult to evaluate patient-centered care outcomes because ‘patient-centered care’ is poorly conceptualized and variably defined (Berwick, 2009); in turn, the lack of a cohesive definition and set of goals makes it difficult to evaluate patient-centered care approaches, particularly when these approaches are enacted for patients with complex needs. Regardless, researchers suggest that patient-centered care means:

Helping patients to be more active in consultations (which) changes centuries of physician-dominated dialogues to those that engage patients as active participants. Training physicians to be more mindful, informative, and empathic transforms their role from one characterized by authority to one that has the goals of partnership, solidarity, empathy, and collaboration. (Epstein & Street, 2011)

How do physicians, nurses and allied health professionals approach care for individuals with DM1, and is it possible to take a ‘patient-centered’ care approach with a chronic disease population that presents with cognitive and behavioral impairments? Since the literature suggests that patients may lack awareness about the significance of their symptoms, I speculate that the complex physical, cognitive and behavioral clinical manifestations of DM1 may influence on-going patient-centered care provision. Others have hypothesized that cognitive impairment may cause patients to be in denial about their diagnosis; consequently, patients may refuse to seek information about their condition, ask for help (Nätterlund, Sjöden & Ahlström, 2001) or miss clinic appointments because they are disinterested in their health (Meola & Sansone, 2007). There is a paucity of research exploring DM1

patients', caregivers' or health care providers' (HCPs) experiences seeking or providing on-going patient-centered care. This is a considerable gap because (1) patients' and caregivers' voices are relatively absent in the literature and therefore we do not know if their needs are being addressed, and (2) it is difficult—if not impossible—to provide, examine or evaluate 'patient-centered' care if patients' experiences are absent.

1.4 Qualitative Explorations of DM1

A review of the literature found few studies that used a qualitative or mixed methods approach to explore adult DM1 (Boström, Alhström & Sunvisson, 2006; Cup et al, 2011; Faulkner & Kingston, 1998; Geirdal, Lund-Peterson & Heiberg, 2014; Heatwole et al 2012; Nätterlund, Sjöden & Ahlström, 2001; Timman, Tibben & Wintzen, 2010); of these, very few occur in a North American context. Moreover, most studies explore individuals living with a range of muscular dystrophies, while only a few are DM1-specific (Cup et al, 2011; Geirdal, Lund-Peterson & Heiberg, 2014; Heatwole et al, 2012; Timman, Tibben & Wintzen, 2010). Regardless, all provide a rich description of symptom impact on patients' and/or caregivers' daily lives; in particular, caregivers describe feeling anxious—and burdened—by their family member's complex needs (Boström, Alhström & Sunvisson, 2006; Cup et al, 2011), and DM1 participants perceive that their altered physical appearance and progressive functional decline impact their sense of identity (Nätterlund, Sjöden & Ahlström, 2001). Furthermore, clinicians and researchers interviewed 20 DM1 patients to elucidate the 'patient reported impact of symptoms in DM1' (PRISM); this exploration was the first phase of a longitudinal project intended to develop patient-centered outcome measures for clinical trials (Heatwole et al, 2012). Findings suggest that patients and HCPs may prioritize different symptoms; that is, participants reported that fatigue had the greatest impact on their lives, yet citations in the literature typically consider myotonia or muscle weakness, not fatigue, to be the primary or characteristic symptoms of DM1. Similarly, Nätterlund, Sjöden and Ahlström (2001) used a descriptive, qualitative approach and interviewed 15 individuals with muscular dystrophy (n =5 DM1) to explore how their lives were impacted by their condition. Findings were condensed to create a 'generic' patient profile that represented

patients' experiences. The DM1 narrative profile suggests that patients' worlds essentially 'shrink' as their disease progresses; that is, patients are no longer able to be employed or to participate in athletic or recreational activities that they once enjoyed. Individuals may also have difficulty making new friends, partly because of a lack of desire to have to continuously explain symptoms like speech impairment and frequent falls (Nätterlund, Sjöden & Ahlström, 2001). Physical symptoms—particularly weakness and fatigue—preclude individuals from engaging in education or employment and restrict their recreational and leisure activities, which resonates with findings from the PRISM study (Heatwole et al, 2012). It is possible that fatigue is widely reported and highly impactful to patients because other symptoms—including muscle weakness, depression or excessive daytime sleepiness—may be labeled and experienced as 'fatigue' (Heatwole et al., 2012). Moreover, patients may lack awareness about their symptoms; an interview study assessing 25 affected females understanding of DM1 found that participants' had variable understanding about their condition, and that their symptomatic knowledge was sometimes informed by their personal experience and family history, not 'book' or medical knowledge. The participants also had misunderstandings about prognosis and genetic information that—in turn—were used to make important health and reproductive decisions (Faulkner & Kingston, 1998). Regardless, weakness and fatigue seem to significantly reduce patients' quality of life. Consequently, findings suggest that these are the symptoms that patients are most concerned about (Heatwole et al, 2012), not those—like cardiac, swallowing or respiratory issues—that clinicians prioritize because of their potential to be life-limiting.

Symptoms and functional decline may negatively impact relationships between patients and caregivers (Cup et al, 2011), and some caregivers describe feeling burdened and anxious about their loved one's deteriorating health (Boström, Ahlström, Sunvisson, 2006). Geirdal, Lund-Peterson and Heiberg (2014) used a mixed methods approach—including quality of life questionnaires and semi-structured interviews—to investigate the quality of life of 13 individuals with DM1 and 8 caregivers. The analysis of the qualitative findings suggests that individuals with DM1 experienced reduced quality of life as a consequence of having to change their expectations; that is, participants acknowledged making difficult reproductive choices, and having reduced levels of

employment and social participation. Individuals with DM1 perceived that others judged them as “lazy” (p. 5) because they were unemployed and limited in their ability to participate in daily activities. In turn, caregivers reported that their quality of life was impacted by isolation and increased responsibilities at home and at work. Life was described as a “roller-coaster”: “Life goes up in the sense that they feel they have the energy, and down when they feel they cannot meet expectations and desires, tasks and challenges...” (p. 5). These findings resonate with results from a hermeneutic study in which five couples living with DM1 were interviewed to explore the effect of DM1 on their relationship (Cup et al., 2011). The couples described that the challenges of living with—and managing—DM1 was akin to the “give and take” of marriage, but that patients’ functional decline required couples’ to renegotiate their roles and reconsider their social and leisure activities. The experience of caregiving may depend on whether it is done out of love or out of a sense of obligation (Boström, Ahlström & Sunvisson, 2006).

However, despite the significant challenges that patients and their caregivers face—and evidence that patients and clinicians prioritize different symptoms--there are no studies that explore DM1 patients perspectives regarding on-going, clinical care. However, a phenomenological approach was used to explore the experiences of patients with various muscular dystrophies (n= 16 DM1) at a multi-disciplinary rehabilitation program in Sweden (Nätterlund & Ahlström, 1999). Patients reported that they felt listened to and understood by expert HCPs who provided them with information about their condition, support and strategies to cope with their illness-related challenges. They also appreciated having the opportunity to interact with others living with similar conditions. These findings resonate with a qualitative study that explored the experiences of adults with neuromuscular disease (n =1 DM1) who attended a neuromuscular rehabilitation centre in the United Kingdom (Hartley, Goodwin & Goldbart, 2011).

1.4.1 Gaps

While the perspectives of patients living with other uncommon neuromuscular conditions like amyotrophic lateral sclerosis and Duchenne muscular dystrophy are well-represented in the literature, there are relatively few studies that use qualitative research to explore

the experiences of individuals living with DM1 (LaDonna, 2011). It is possible that researchers and clinicians do not use qualitative methodologies and methods with DM1 patients because of their perception that individuals with DM1 are likely to have significant cognitive or behavioral impairments. For example, researchers who explored the illness experience of patients living with muscular dystrophy excluded some individuals with DM1 because of difficulty understanding their speech (due to oropharyngeal muscle weakness) and cognitive impairment (Nätterlund, Sjöden & Ahlström, 2001). Furthermore, the authors noted “these conditions prevented them from reflecting deeper upon their situation” (Nätterlund, Sjöden & Ahlström, 2001, p. 790). As a result of limited qualitative research exploring DM1-affected individuals perspectives about their health care, we do not know if the voices of those with DM1 are being heard by clinicians and researchers, or if their clinical and psychosocial needs are being met. Moreover, no studies provide an exploration of health care providers’ experiences providing on-going care for individuals along the DM1 disease trajectory. Therefore, we know little about clinicians’ approaches, or whether HCPs perceive that they are meeting patients’ needs. The multi-system nature of DM1 makes it an important condition to explore because findings may have implications for a host of complex, chronic conditions that present with multiple co-morbidities.

1.4.2 Purpose

The purpose of this research was to explore patients’ and caregivers’ experiences of living with DM1, including their health care expectations and motivations for maintaining on-going clinical care in an outpatient neurology clinic. This research also sought to explore HCPs perspectives about care provision for persons living with DM1. I chose a qualitative research approach to: (1) add patients’ and caregivers’ voices to the neuromuscular disease literature, (2) explore on-going care provision for individuals with DM1, and (3) probe whether patients’ and caregivers’ clinical and psychosocial needs are being met. Finally, I was interested in exploring what patient-centered care provision ‘looks like’— if it is feasible, and if it occurs—in DM1 clinical care. Therefore, individuals with DM1 and their caregivers were recruited from a neuromuscular clinic at one academic centre in Ontario, Canada. To facilitate a deeper exploration of patient-

centered care provision in chronic neurological conditions, I invited patients with Huntington's disease (HD), their caregivers and health care providers to participate in a study exploring motivations for clinic attendance. HD is an uncommon neurological movement disorder characterized by involuntary movements, progressive physical and cognitive decline, and significant psychiatric manifestations including depression, anxiety, obsessions, and psychosis (Roos, 2010; Sturrock & Leavitt, 2010). DM1 and HD have a number of similarities: (1) both conditions are autosomal dominantly inherited and characterized by anticipation, (2) they have highly variably clinical manifestations that include motor, cognitive and behavioral features, (3) present with complex biopsychosocial issues, and (4) there are no treatments to cure, slow, or reverse neurodegeneration. The purpose of adding an exploration of care provision in HD was not to compare the two populations; instead, the goal was to add breadth and depth to how care at one academic centre is provided for patients with chronic neurological disease who present with cognitive and behavioral impairments.

1.5 Research Summary

This research includes three studies that produced five manuscripts (Figure 1; Table 1). The perspectives of 49 participants are included in this research; details about the research setting and study sample are described in the relevant chapters. Twenty-one patients including 13 individuals with adult-onset DM1 and 9 individuals with HD, 16 caregivers (n= 8 DM1) and 11 HCPs participated. The HCPs included 5 neurologists, two specialist physicians including a psychiatrist and a respirologist, a nurse, two social workers and a physiotherapist. Of note, one individual with DM1 participated in two studies.

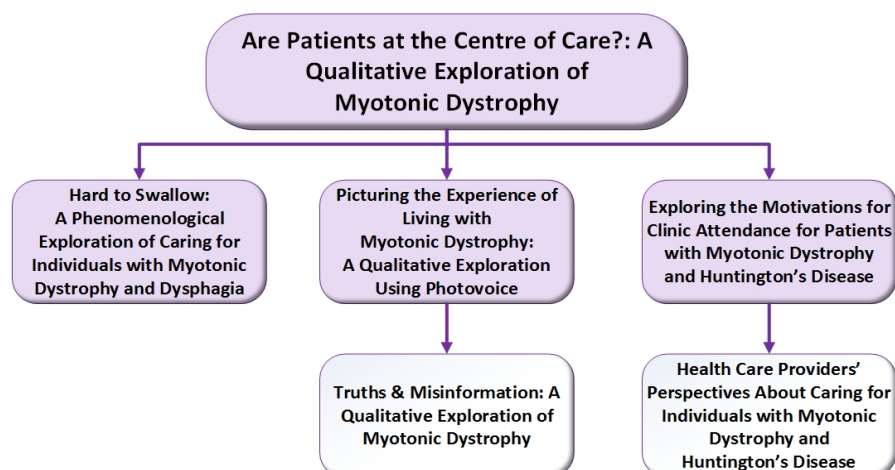


Figure 1-1 : Dissertation Research

1.6 Chapter III: Hard to Swallow: A Phenomenological Exploration of Caring for Individuals with Myotonic Dystrophy and Dysphagia.

The literature suggests that cardiac, respiratory and swallowing dysfunction are the most life-threatening symptoms in DM1—and the symptoms that clinicians may preferentially monitor—because they have the propensity to cause serious complications including sudden death (Garrett, DuBose, Jackson, & Norman, 1969). Dysphagia should be one symptom that clinicians, patients and caregivers can proactively address through education, evaluation and dietary management. The literature suggests that caregivers for patients with other chronic illnesses that present with dysphagia are responsible for managing their loved one's diet and responding to choking emergencies (Johansson & Johansson, 2009), therefore, the purpose of this study was to explore the experience of caring for an individual with DM1 and known dysphagia. Phenomenology—a qualitative research methodology that explores lived experience (van Manen, 1990)—was used, and individuals with DM1 and dysphagia (by physician report) were approached by their health care provider and asked to identify a caregiver and provide him or her with a letter of information about the study. Six caregivers consented to participate in interviews about their caregiving experiences. The interview transcripts were transcribed verbatim and analyzed by considering participants' experiences in the context of their “lived body”, “lived time”, “lived space” and “lived relations”.

1.7 Chapter IV: Picturing the Experience of Living with Myotonic Dystrophy: A Qualitative Exploration Using Photovoice.

Following the exploration of the caregivers' perspectives, I was interested in exploring patients' experiences of living with DM1. Since the literature suggests that patients may have cognitive, behavioral and speech impairments, an innovative approach for conducting research with this population was warranted. Therefore, photovoice—a novel qualitative visual research methodology that was developed to engage participants with low literacy as research collaborators (Wang & Burris, 1994; 1997)—was used to explore participants' illness experiences and the barriers and facilitators to living successfully

with DM1. Nine participants were given digital cameras and asked to “take pictures of what it is like to live with DM1”; in turn, participants’ photographs stimulated individual interviews and focus group discussions. The purpose of this study was (1) to use an innovative qualitative research methodology to explore the experiences of living with DM1, (2) to identify barriers and facilitators to living successfully with DM1, and (3) to assess if photovoice is a useful methodology to use to conduct research in this population.

1.8 Chapter V: Truths and Misinformation: A Qualitative Exploration of Myotonic Dystrophy Type 1.

While analyzing the data from *Picturing the Experience*, it became evident that participants had variable knowledge about their condition, and “Truths and Misinformation” – a theme that was identified during data analysis for the *Picturing the Experience* study—warranted a deeper exploration. Therefore, I worked with two clinicians to conduct a content analysis of this theme to examine how participants talk about—and make sense of—their condition. Transcripts were re-read and re-coded to determine patients’ understanding of DM1—including their awareness or misinformation about symptoms, treatment and prognosis; in turn frequently occurring codes were collapsed into themes and categories, and supporting quotations were extracted from the data set.

1.9 Chapter VI: “[The Neurologist] is Throwing you a Raft”: Exploring Motivations for On-going Clinic Attendance for Individuals Living with Chronic, Progressive and Life-limiting Neurological Conditions.

The literature suggests that individuals with DM1 and HD may lack awareness about their symptoms (Boström & Ahlström, 2005; Hoth et al, 2007) and our findings resonate with previous literature suggesting that individuals with DM1 have variable knowledge about their condition (Laberge et al, 2010; Faulkner & Kingston, 1998). Given the lack of disease-halting or curative treatment options for DM1 or HD—coupled with the potential that patients are disinterested in their health and unaware of problematic symptoms—it is unknown why individuals with these conditions maintain regular, on-going follow-up at

an outpatient neurology clinic. Therefore, this research sought to elucidate patients' and caregivers' motivations for clinic attendance and their health care expectations, and HCPs perspectives about care provision for these conditions.

Constructivist grounded theory (CGT)—a qualitative research methodology that studies basic social processes to generate an explanatory theory—informed the iterative data collection and analysis process (Charmaz, 2006; 2014). 14 patients (n=5 DM1), 10 caregivers (n=2 DM1), and 11 HCPs (n=5 neurologists; 2 specialist physicians; a nurse; two social workers and a physiotherapist) participated in semi-structured interviews that were transcribed verbatim and coded using words or phrases that represented the participants' experiences. DM1 and HD data were not analyzed separately, and the themes and categories represent the perspectives of patients, caregivers, and HCPs from both groups.

1.10 Chapter VII: “We Like to Think We’re Making a Difference”: Health Care Providers’ Perspectives About Caring for Individuals with Myotonic Dystrophy and Huntington’s Disease.

The HCPs' perspectives about motivations for clinic attendance for individuals with DM1 and HD warranted a deeper exploration and were presented in a separate manuscript; this analytical project used the same data collection and analysis procedures as the previous study.

1.11 Acknowledging the Boundaries

In summary, my doctoral research has three main goals: (1) to use qualitative research methodologies and methods to explore the experiences of patients, caregivers and health care providers who live with—or provide care for—individuals living with myotonic dystrophy, (2) to add patients' and caregivers' voices to the largely biomedical neuromuscular literature, and (3) to explore what DM1 care ‘looks like’ at one academic medical centre in Ontario, Canada. The purpose of this research, however, is not to evaluate patient-centered care in DM1 because: (1) there is not a uniform definition or conceptualization of patient-centered care, therefore rendering it impossible to evaluate,

(2) the literature does not yet report whether ‘patient-centered care’ is implemented, feasible or achievable in DM1 care, and (3) DM1 patients’ and caregivers’ perspectives are under-represented in the literature suggesting that the ‘patient’ is not currently present in conversations about DM1 ‘patient-centered care’. Rather, the intention of this research program is to contribute to a scholarly conversation about patient-centered care provision in DM1.

Title	Authors	Journal	Status
<i>Hard to Swallow: A Phenomenological Exploration of Caring for Individuals with DM1 and Dysphagia</i>	K.A. LaDonna, W.J. Koopman, S.L. Ray & S.L. Venance	Journal of Neuroscience Nursing	Accepted: May, 2014
<i>Picturing the Experience of Living with Myotonic Dystrophy: A Qualitative Exploration Using Photovoice</i>	K.A. LaDonna & S.L. Venance	Journal of Neuroscience Nursing	Accepted, pending revisions: June, 2014 Revisions submitted: September, 2014
<i>Truths & Misinformation: A Qualitative Exploration of Myotonic Dystrophy</i>	K.A. LaDonna, A. Ghavanini & S.L. Venance	Canadian Journal of Neurological Sciences	Submitted; April, 2014 Invitation for revision: October, 2014 Revisions submitted: November, 2014
<i>Exploring Motivations for On-Going Clinic Attendance for Individuals Living with Myotonic Dystrophy and Huntington's Disease</i>	K.A. LaDonna, C.J. Watling, S.L. Ray & S.L. Venance	Qualitative Health Research	For submission: January, 2015
<i>"We Like to Think We're Making a Difference": Health Care Providers' Perspectives About Caring for Individuals with Myotonic Dystrophy and Huntington's Disease</i>	K.A. LaDonna, C.J. Watling, S.L. Ray, C. Piechowicz & S.L. Venance	Qualitative Health Research	Submitted: November, 2014

Table 1-1: Research Summary

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Chapter 2

2 Methodologies and Methods

This chapter describes the background and theoretical underpinnings of phenomenology, photovoice and grounded theory, and describes the methods associated with these methodologies. Methodologies and methods are distinct; methodologies are guiding frameworks that are based on conceptual, theoretical, or philosophical foundations, while methods are the tools used to conduct research within the framework of the chosen methodology (Finlay, 2006). For instance, an in-depth or semi-structured interview is a frequently used research method for many qualitative methodologies like phenomenology, photovoice or grounded theory (van Manen, 1990; Wang & Burris, 1994; 1997; Charmaz, 2006; 2014). The researcher's choice of methodology and method generally reflects his or her research question and location along the spectrum of qualitative research paradigms. Therefore, a researcher located within the positivist research paradigm might choose to combine statistical analysis with a traditional grounded theory approach (Glaser & Strauss, 1967), while critical theorists—who are located on the opposite end of the spectrum—would be more likely to use methodologies and methods based on participatory action research or feminist theory (Finlay, 2006). I align myself within the constructivist paradigm because I do not believe that there is an 'objective truth' that can be discovered; rather, I believe that truth is subjective and that meaning is influenced—and therefore co-constructed by—researchers' and participants' experiences. Consequently, I chose three qualitative research methodologies—phenomenology (van Manen, 1990), photovoice (Wang & Burris, 1994; 1997) and constructivist grounded theory (Charmaz, 2006; 2014)—that recognize how individual experience and the research setting may influence data collection, analysis and findings. Following a description of the methodologies, considerations for ensuring the trustworthiness of qualitative research data will be discussed.

2.1 Participants and Research Setting

The specific data collection and analysis procedures for each study will be described in Chapters III-VII. However, all research participants were recruited from one academic medical centre in Ontario, Canada. At this centre, one neuromuscular physician—with clinical support from a nurse practitioner and/or medical students, residents or fellows—provides care for most DM1 patients within the specialist’s general adult muscle disease clinic. Patients are typically followed every six months to two years (depending on need), and the clinic staff has access to other specialist physicians and allied health professionals including a social worker, dietician, occupational therapist and a physiotherapist on a referral basis. However, the allied health professionals typically only see DM1 patients on a one-time referral basis for a specific issue. Huntington’s disease patients (who were explored in Chapters VI and VII) may be seen by any of the four movement disorder specialists at the centre, but most are followed in a dedicated Huntington’s disease clinic that is held once a month. At the HD clinic, a neurologist, psychiatrist and social worker typically conduct clinic visits concurrently. At both clinics, caregivers and other family members may also access clinic staff.

2.2 Chapter III: Hard to Swallow: A Phenomenological Exploration of the Experience of Caring for Individuals with Myotonic Dystrophy (DM1) and Dysphagia

The purpose of this study was to explore the lived experience of caregivers for individuals with DM1 and swallowing dysfunction. Participants were posed with an open-ended question about the experience of caring for someone with DM1 and dysphagia. To follow-up, participants were asked to describe their caregiving duties, ideas about strategies for safe swallowing, their opinions about their loved one’s perceptions of DM1 and swallowing dysfunction, and the impact of DM1 on their lives. A phenomenological research approach was chosen to explore the lived experiences of caregivers for individuals with DM1.

2.2.1 Methodology: Phenomenology

Phenomenology is both a philosophy and a research methodology that encompasses a variety of European and North American traditions (Dowling, 2007; Earle, 2010). The philosophy of phenomenology was developed in early 20th century Germany to discuss the idea of truth (Dowling, 2007), and the ‘father of phenomenology’, Edmund Husserl, devoted much of his work to understanding “what is real and valid, what constitutes evidence, and what is the relationship between the knower and the known” (Todres, 2005, p. 104). In particular, Husserl (1970) was interested in the lifeworld (*Lebenswelt*) and essences, and he sought to understand human experience by examining things as they appear. Husserl was interested in intentionality, reduction and the constitution of meaning - concepts that continue to influence the various and evolving phenomenological traditions (Earle, 2010). Intentionality invokes the ability of humans to be aware of, and to be able to reason and communicate about objects in their lifeworld (Earle, 2010). For Husserl, this understanding should be pre-reflexive. To do this, Husserl proposed that individuals engage in phenomenological reduction to ‘bracket’ or set aside their knowledge and their preconceptions about an experience; that is, individuals should attempt to understand a phenomenon without a cultural context (Dowling, 2007), preconceived theoretical ideas (van Manen, 1997), or interpretations (Dowling, 2007). Through intentionality and reduction, individuals are able to identify the essences and meaning of an experience (Earle, 2010).

Husserl was primarily interested in the description of experiences (Earle, 2010). However, the writings of Martin Heidegger, a student—and later critic—of Husserl shifted phenomenological inquiry from descriptive to interpretive (Dowling, 2007). Heidegger’s main interest surrounded the meaning of Being, or the individual’s presence in the world, and he differed from Husserl in his belief that to understand an experience is more important than describing it (Dowling, 2007). Heidegger’s *Being and Time* (1927) focuses on the meaning of being in the world (*Dasein*) (Earle, 2010). For Heidegger, exploring existential concepts like temporality (i.e., the past, present and future) is essential for understanding being, and he criticized Husserl’s ideas of intentionality and reduction (Dowling, 2007; Earle, 2010). In particular, Heidegger believed that it is

impossible to set aside one's assumptions and pre-conceptions about an experience. Instead, Heidegger proposed the idea of the hermeneutic circle in which individuals examine the 'parts and whole' of a phenomenon by continuously examining and comparing their pre-suppositions and understandings of an experience with the unfolding essence of being (Earle, 2010).

French philosopher Maurice Merleau-Ponty's work evolved from the writings of Husserl and Heidegger, but he emphasized the "primacy of perception" (Dowling, 2007, p. 134). Merleau-Ponty was interested in viewing experiences pre-reflexively, not using pre-defined categories, and he was particularly interested in exploring the existentials of *lived space*, *lived time*, *lived human relation*, and *lived body* (Dowling, 2007). The next generation—or New Phenomenologists—follow the tenets of either interpretive (e.g. Heidegger or Merleau-Ponty) or descriptive phenomenology (Husserl). However, Max van Manen (1990) incorporates both interpretive and descriptive phenomenology in his version of the methodology. Further, van Manen views phenomenological inquiry and analysis through the lens of the four existentials of being in the world, and also considers the role of the researcher in data collection and analysis (van Manen, 1990).

In her monograph titled *Illness: The Cry of the Flesh* (2008), philosopher Havi Carel argues that the naturalistic approach to illness is insufficient for understanding the life changing impact that chronic illness has on an individual. Thus, Carel makes a strong argument for using phenomenology to study the experiences of individuals living with chronic illness. In particular, Carel believes that "phenomenology does not deny the importance of the physiological description or of the clinical interventions offered by current mainstream medicine. It does, though, propose to augment this approach to illness by emphasizing the importance of the first person experience" (2008, p. 8).

2.2.2 Method: Van Manen

One of the difficulties facing novice researchers undertaking a phenomenological exploration is that there is no uniform method to collect or analyze data (Caelli, 2000). Van Manen (1997), however, articulates an approach to human science research that engages researchers in six research components: (1) to explore a phenomenon of interest;

(2) explore this experience as lived rather than as it is conceptualized; (3) reflection on essential themes; (4) describing a phenomenon through the art of writing; (5) remaining orientated to the phenomenon, and (6) being mindful of the ‘parts and wholes’ of the research context.

Van Manen describes several possibilities for data collection and analysis; however he cautions against using these suggestions as a ‘prescription’ for phenomenological work. Instead, he encourages researchers to approach the question creatively.

Phenomenological research views the lifeworld as the “source and object” of inquiry; therefore, the “nature of data” (van Manen, 1997, p. 53) can take many forms including the researcher’s personal experience, research participants’ descriptions, observation, biography, literature and art. In particular, interviews with research participants may serve two purposes: (1) to explore a participant’s experiences to gain a deeper understanding of a phenomenon, or (2) to embark on a conversation with a participant to explore the meaning of an experience (van Manen, 1997). Van Manen (1997) suggests asking participants a concrete question about a phenomenon, then encouraging them to describe anecdotes, incidents, and stories regarding this experience. It is not necessary to ask a litany of questions; rather, participants should be given the freedom to articulate their experience. The researcher may then use probes to gain a deeper understanding of the participant’s narrative.

Once data has been collected, the researcher reflects on the findings to try to understand their meaning. Van Manen (1997) distinguishes between the researcher’s pre-reflective and reflective understanding of an experience; that is, one may be able to describe that life is regulated by time, but have difficulty discussing what time actually means or ‘is’. Therefore, “the insight into the essence of a phenomenon involves a process of reflectively appropriating, of clarifying and of making explicit the structure of meaning of the lived experience” (Van Manen, 1997, p. 77). This may be done in a variety of ways including thematic analysis. Thematic analysis may be explored using a holistic, selective, then detailed approach (van Manen, 1997). During the holistic or sententious approach, the researcher reads the text as a whole, then writes a sentence or phrase that captures the essence of the narrative. Next, the researcher may highlight statements or

phrases that exemplify the experience as lived. Finally, the researcher reads each sentence or sentence cluster in detail to determine what they ‘reveal’ about the phenomenon being explored. Once a list of themes has been drafted, the researcher may return to the participants to ask them to reflect on the findings to ensure that the themes resonate with their experience. Therefore, participants become active collaborators in interpretation and analysis.

Van Manen suggests using the lifeworld existentials of *corporeality*, *spatiality*, *temporality* and *relationality* as guides for hermeneutic phenomenological reflection. These four existentials may guide the way humans experience the world (van Manen, 1997). Corporeality refers to the idea that humans are always “bodily in the world” (p. 103). Van Manen describes how human bodies may unconsciously ‘reveal’ or ‘conceal’ aspects of the human experience; that is, he suggests that one may behave awkwardly when looked at by another critically, or conversely, develop grace when gazed at admiringly. Spatiality refers to “felt space” or “the world or landscape in which human beings move and find themselves at home” (p. 102). How one experiences a physical space gives meaning to a phenomenon, and influences how an individual feels in certain situations. For example, one may feel small in a large, open space, or feel lost or vulnerable in an unfamiliar space (van Manen, 1997). Van Manen argues that, in a sense, humans become the space they inhabit. Temporality, or lived time, is subjective time rather than ‘clock’ time. The idea that ‘time flies when you’re having fun’ speaks to temporality, and the past, present and future make up an individual’s “temporal landscape” (van Manen, 1997, p. 104). Finally, relationality refers to the lived relation individuals have with each other in a shared space. Relationality may confirm—or not— notions one has of others.

Writing is an intrinsic part of the phenomenological research process that should occur simultaneously with data collection and analysis, not simply as a means to report findings (van Manen, 1997). In order to balance the ‘parts and wholes’ of the research context, researchers should have an understanding of how the study will be conducted and how the text may be structured, but that researchers are free to flesh out these details and make decisions as the study unfolds (Earle, 2010; Van Manen, 1997).

2.3 Chapter IV: Picturing the Experience of Living with Myotonic Dystrophy (DM1): A Qualitative Exploration Using Photovoice / Chapter V: Truths and Misinformation: A Qualitative Exploration of Myotonic Dystrophy

The purpose of the study described in Chapter IV was to use photovoice—an innovative qualitative research approach—to explore the experience of living with DM1. Nine participants were given a digital camera and asked to take pictures of ‘what it is like to live with DM1’. The goals of this study were to elucidate individual experience about DM1, to identify barriers and facilitators to living successfully with DM1, and to assess whether photovoice is a useful methodology for conducting research with this population. One theme identified during data analysis was titled *DM1: Truths & Misinformation*, and pertained to affected individuals understanding or misperceptions about DM1. The purpose of the paper described in Chapter V was to provide a deeper exploration of this theme and to examine patients’ variable understanding of their condition, and the potential implications of ‘truths & misinformation’ for patient-centered care provision. Content analysis (Miles & Huberman, 1994) was used in conjunction with participant-directed analysis to analyze the data for both studies.

2.3.1 Methodology: Photovoice

There seems to be a debate amongst researchers as to whether photovoice is a methodology, a method, or both. A scan of titles produced during a literature search illustrates this confusion. That is, titles—even by the author who conceptualized photovoice—variably refer to photovoice as a method or a methodology (Wang & Burris, 1997; Baker & Wang, 2006). I argue that Wang and Burris (1994; 1997) originally intended photovoice to be a method for Participatory Action Research (PAR), but that photovoice has since evolved into its own distinct methodology, yet continues to be used as a data collection method for other methodological approaches (Plunkett, Liepert & Ray, 2013). A scoping review of the photovoice literature found that 55% of the 191

studies use photovoice as the sole methodology and method for data collection and analysis, while 45% use photovoice as method for other qualitative or quantitative methodological approaches (Lal, Jarus & Suto, 2012).

Regardless, photovoice is rooted in—and shares many of the philosophical and theoretical conceptualizations as—PAR. Both methodologies are located within the critical research paradigm and are a “systematic investigation, with the collaboration of those affected by the issue being studied, for the purposes of education and taking action or effecting social change” (cited in Minkler, 2000). Social psychologist Kurt Lewin (1946; 1952) coined the phrase “action research” to describe a cyclical research process that includes planning, action, observation and evaluation. Both PAR and photovoice were influenced by Paulo Friere’s work on critical consciousness (Freire, 1970; Fals-Borda, 1987; Wang & Burris, 1994; 1997); in particular, Freire was an educator and proponent of problem-focused education and believed that a student can be an educator while the teacher can become a student. In turn, a problem or opportunity for change is identified by a group of community members who then work together with a researcher to determine where and how to make improvements (McTaggart, 1991; Minkler, 2000). “Put simply, action research is the way groups of people can organize the conditions under which they can learn from their experiences and make this experience accessible to others” (McTaggart, 1991, p. 170). PAR and photovoice distinctly differ from research done to or on research participants (McTaggart, 1991). Instead, PAR and photovoice strive to include participants in all stages of the research process, from the conceptualization of the question, to data collection and analysis, and finally to dissemination of the findings and decision-making about actions to change or improve community outcomes (McTaggart, 1991). In PAR and photovoice studies, participants are viewed as experts about their experiences and community concerns, and are engaged as active collaborators on important research decisions (Wang & Burris, 1994; 1997). Attention to power differentials is an important component of both methodologies, particularly because there may be a real or perceived power imbalance between academic researchers and participants (Wang & Burris, 1994; 1997).

However, photovoice –which was developed by Wang and Burris (1994; 1997) as a

needs assessment tool to explore the reproductive health issues of rural Chinese women—has combined aspects of a number of methodologies and theories to form its own distinct methodology with innovative data collection and analysis procedures. Originally titled photo novella, Wang and Burris (1994) created their method based on concepts from educator Paulo Freire (1970), feminist theory, and documentary photography. In particular, they emphasize the use of female intellect and experience to inspire problem-posing education and individual development for personal and social change. Ultimately, Wang and Burris (1994) believe that the visual image is a powerful conduit for discussion and change because photographs provide evidence and validation of community concerns.

There are three major goals of photovoice: (1) to have community members record their perceptions of the strengths and weaknesses of their surroundings, (2) to use photography to encourage dialogue among community members, and (3) to disseminate this information to policy makers (Wang & Burris, 1997). Arguably, the most important aspect of photovoice as a needs assessment tool is that participants become advocates for change (Wang & Burris, 1997), and to empower participants by putting “cameras directly in the hands of people who otherwise would not have access, and allows them to be recorders, and potential catalysts, in their own communities” (Wang & Burris, 1994). Photovoice adds humanity to data (Wang, Burris, & Ping, 1996) thus, participants, not researchers, determine what is important to document and discuss with peers, academic researchers, and policy makers.

Photovoice has been used with adult and children in a variety of contexts including explorations of homelessness (Aitken & Wingate, 1993; Fortin, 2014), motherhood (Booth, 2003) and child and adolescent programs (Berinstein & Magalhaes, 2009). Additionally, it has been widely used in health research to explore a variety of chronic illnesses including HIV/AIDS (Kubicek et al, 2012; Moletsane et al, 2007), cancer (Lopez et al, 2005), chronic pain (Baker & Wang, 2006) and mental illness (Andonian, 2010); moreover, it is becoming an important research methodology to explore patients with complex needs including speech, cognitive and behavioral impairments. In particular, it has been successfully used to explore chronic neurological disease

populations including spinal cord injury (Newman, 2009; 2010), traumatic brain injury (Lorenz & Kolb, 2009), dementia (Genoe & Dupuis, 2013), stroke (Levin et al, 2007), Alzheimer's disease (Wiersma, 2008; 2011), and with Huntington's disease caregivers (Aubeeluck & Buchanan, 2006).

2.3.2 The Photovoice Method: Data Collection and Analysis

While there are no set guidelines for a photovoice study, descriptions from Wang and Burris (1994) suggest that a photovoice study might include an orientation session, focus groups and/or individual interviews, and a forum to disseminate findings. Camera orientation sessions should be tailored to the culture of the community, and ethics, power regarding data ownership and dissemination, camera operation, and guiding principles about the project are typically discussed (Wang & Burris, 1997). In particular, the authors suggest that facilitators guide participants about the appropriate and ethical way to approach others to be in pictures, the ethics of taking pictures without the knowledge of others, the criteria to be used when analyzing photographs, and the personal and societal implications of taking pictures and sharing them publicly.

While Wang and Burris (1997) suggest that the discussion of camera use should be minimal to prevent "stifling creativity" (p. 378), it is important to ensure that participants are comfortable operating the camera. A significant portion of the camera orientation session should be devoted to discussing ethics, power, and data ownership. Participants should be reminded that confidentiality is of utmost importance, and that anything shared during interviews or focus groups will be de-identified prior to dissemination. Further, participant names or faces (unless specific permission is granted) will never be published. Importantly, the camera orientation session might include a discussion about academia and the desire for researchers to publish findings or present at conferences. However, the participants should be free to determine if and how they wish to disseminate findings.

An individual interview is an opportunity for participants to discuss as many of their photographs as they wish, and to offer insight that they may be unwilling to share in a group setting. It is important that participants guide the discussion by choosing and discussing the implications of their photographs. Participants are also encouraged to

keep a journal or log book of what they photographed and why. It is also important to note what a participant was either unable to, or chose not to, photograph. Log books may serve as guideposts for the interview discussion and become an important part of data analysis. It may be advantageous at this stage to ask participants about the benefits and challenges associated with participating in the project. Another strategy to facilitate the participants' participation in data collection and analysis might be to ask participants to title each of their photographs.

There is no singular method for analyzing photovoice data, and authors (Olliffe & Bottorff, 2007; Olliffe, Bottorff, Kelly, & Halpin, 2008) have chosen a variety of analytical approaches. However, Wang and Burris (1997), and Wang and Redwood-Jones (2001) describe their data analysis methods. Wang and Burris (1997) offer guidelines for data analysis, and they reinforce that participants are an integral part of this process. In particular, the authors discuss a three-stage approach to analysis in which participants select, contextualize, and codify the data. By selecting which photographs to discuss, the participants guide the first step of data analysis. Participants then contextualize their photographs by describing why they chose to capture certain objects, people or settings, and therefore give meaning to their images. This part of the analysis typically occurs during the individual interview or focus group session when participants have the opportunity to discuss how their photographs relate to community issues and personal experiences. Finally, participants codify the photographs through discussion of emerging themes, theories, and issues embedded in their photographs and collective experience. Participants therefore engage with researchers to analyze the data; in turn, researchers use their own experiences and expertise to co-construct findings.

In addition, several studies have used content analysis (Hergenrather, Rhodes, & Clark, 2006; Thompson, et al., 2008) or tenets from grounded theory (Lopez, Eng, Randall-David, & Robinson, 2005) to analyze data. Lopez et al. (2005) chose to blend grounded theory and photovoice to “provide the means for participants to move beyond merely reporting results to policy and decision makers to suggesting strategies and participating in developing interventions tailored to specific conditions of their social context” (p. 101).

2.3.3 Ethical Considerations

Wang and Redwood-Jones (2001) write extensively about the ethical considerations of the photovoice method. In particular, the authors discuss privacy laws and note that intrusion into public spaces, disclosure of embarrassing issues, being placed in a false light, and using a person's likeness without compensation are serious concerns that must be addressed throughout the process. In terms of a photovoice project, participants must understand that photographers create meaning by capturing and interpreting a subject or location (Wang & Redwood-Jones, 2001). Also, the authors discuss the importance of ensuring participants that they own their photographs by giving them the negatives of all of their pictures. Also, when possible, subjects of photographs should be provided compensation if their images are disseminated publicly. In particular, camera orientation facilitators should ask participants to reflect on the responsibilities of using a camera, how to respectfully approach someone about having his or her picture taken, and how to minimize risks to self and others (Wang, 2004 #38).

Informed consent is an integral part of the photovoice project. Participants should be given a consent approved by a university ethics board describing the purpose of the study, their rights and responsibilities, and their willingness to allow their images to enter the public domain. While participants may not be required to obtain consent when taking a group photograph in a public setting where individuals are non-identifiable (Wang & Redwood-Jones, 2001), participants should provide their photography subjects with a separate consent outlining the study and the rights of those having their pictures taken. In particular, all identifiable subjects should understand how and why their image will be used, and that no image shall be published or disseminated publicly without their written consent. As with all research studies, consent is a process that should be revisited throughout the project.

2.3.4 Considerations and Benefits

Since photovoice is an innovative and relatively new methodology, the conceptual and theoretical underpinnings need to be critically examined (Guillemin & Drew, 2010),

particularly since there is considerable debate whether photovoice is a PAR method or its own distinct methodology. Also, using cameras to document experiences may be a novel concept that may be challenging for some participants, and in-depth discussions about the purpose of the research and instructions about how to use the camera are crucial and should be re-visited throughout the research process (Guillemin & Drew, 2010). Further, Foster-Fishman, Nowell, and Deacon (2005) discuss the importance of understanding the impact that a photovoice project may have on both the participants and their community. Thus, these authors argue that it is essential to ask participants to not only discuss their photographs, but to ask them about their experience using the method, and how the project impacted them. Potential impacts of participation may include enhanced self-competence, greater awareness of their environment, and the potential for photovoice projects to result in increased resources for social and political action (Foster-Fishman, Nowell, Deacon, Nievar, & McCann, 2005). However, one must be cognizant of the negatives that may be encountered by being both the participant in, and the subject of, publicly disseminated photovoice data. In particular, participation may have negative political consequences (Wang & Burris, 1994), or participants may fear public disclosure of personal health issues. Therefore, questions of who owns the photos and data are important to consider and communicate with participants: “This is particularly pertinent during analysis, when considering questions of who had control over the camera and the image-making moment, and whose understanding or experience is being represented in the image; the commentary of participants is crucial in understanding this element of construction of the visual story” (Guillemin & Drew, 2010). It is difficult—if not impossible—to know the extent to which the researcher may influence what and how participants choose to capture their experiences. Guillemin and Drew (2010) are interested in the concept of the ‘audience’ for a photovoice project—and the role of the researcher in particular-- and suggest that ‘audience’ is a concept that needs to be explored.

2.4 Chapter VI: Understanding Motivations for Clinic Attendance and Expectations for Care for Individuals with Myotonic Dystrophy (DM1) and Huntington’s

Disease (HD) / Chapter VII: “We Like to Think We’re Making a Difference”

The studies that will be presented in Chapters VI and VII sought to elucidate patients’, caregivers’ and health care providers’ perceptions about the motivations for patients with DM1 and HD to maintain regular follow-up at an outpatient neurology clinic despite the lack of curative treatments. Participants were asked to describe a ‘typical’ clinic visit, and to reflect on their expectations of—or approaches to—health care. Constructivist grounded theory (Charmaz 2006) guided the iterative data collection and analysis process. A deeper exploration of HCPs perspectives about providing care for individuals with DM1 and HD is explored in Chapter VII.

2.4.1 Methodology: Constructivist Grounded Theory

Grounded theory is an appropriate methodology to use to study the process of motivation and decision making for individuals with a chronic neurological illness who choose to receive regular care in an outpatient clinic despite the lack of a cure or treatment to slow the progression of their disease. Not only is grounded theory a relevant methodology to use when little is known about a topic (Stanley, 2006), but Charmaz (1990) argues that research using grounded theory may offer physicians an exploration of patients’ views that cannot be gained during a clinic visit. In particular, study findings may encourage better communication between doctors and patients, and focus attention on concerns that patients identify (Charmaz, 1990). Constructivist grounded theory (Charmaz, 2006), the proposed methodology for this project, is a version of grounded theory that considers the interplay of the researcher and the research participant in co-constructing the data. However, prior to proposing a constructivist grounded theory approach, it is important to understand the evolution of the grounded theory methodology.

2.4.1.1 Methodology

Grounded theory was first introduced in the 1960s by sociologists Barney Glaser and Anselm Strauss as a methodology to generate theory that is grounded in data. The

ultimate purpose of grounded theory is to use induction, deduction, and verification to develop a theory about a social phenomenon (Schwandt, 2007). In particular, practitioners of grounded theory use their preliminary data analysis to guide data collection, and iterative data collection and analysis inform emerging theory (Charmaz, 2003).

Glaser and Strauss' book *The Discovery of Grounded Theory* (1967) was written at a time when qualitative research was losing prestige and quantitative methods were gaining momentum. Following World War II, quantitative research that gathered evidence to test theories gained momentum, thus consigning qualitative methods to data gathering or exploratory tools for survey development. In other words, qualitative research was useful for generating hypotheses, but then quantitative research would "take over" to test theories (Glaser & Strauss, 1967, 16). Glaser and Strauss (1967) responded by arguing that both quantitative and qualitative data are useful and important for verifying or generating theory, and they proposed a methodology in which "qualitative analysis had its own logic and could generate theory" (Charmaz, 2006, p. 5). Charmaz (2008) argues that Glaser and Strauss' work legitimized qualitative research. According to Glaser and Strauss (1967), a grounded theory study should include the following: an iterative process of data collection and analysis, codes and categories that are grounded in the data, a constant comparative method, theory development at each stage of research, memo writing, theoretical sampling, and a literature review that is conducted after the analysis. However, the guidelines and methods for study conduct may be flexible and emerge throughout the process (Charmaz, 2008).

According to Charmaz (2006), Glaser and Strauss each brought experiences from their respective training at Columbia University and the Chicago school culture while developing grounded theory. In particular, Glaser wanted researchers to develop middle-range theories, and he "imbued grounded theory with dispassionate empiricism, rigorous codified methods, emphasis on emergent discoveries, and its somewhat ambiguous specialized language that echoes quantitative methods" (p. 7). Strauss emphasized the importance of process, problem-solving, the study of action, and subjective meanings. Further, the grounded theory of Glaser and Strauss is influenced by symbolic

interactionism. Symbolic interactionism, both a theory about human behavior and an approach for studying it (Annells, 1996), defines an individual as being socially constructed, and argues that individuals use social interaction processes to navigate their world (Stanley, 2006). This definition is based on the work of social psychologist George Herbert Mead (1962) who proposed that individuals define themselves through social interactions and perspectives. In other words, “humans come to understand social definitions through a socializing process” (Annells, 1996, p. 381). Blumer (1969) extended Mead’s work by coining the term ‘symbolic interactionism’ and proposing three basic tenets about the concept: (1) the meanings that individuals ascribe to others, objects, or situations will determine their actions towards them, (2) meanings stem from social interactions, and (3) individuals undertake an interpretive process to determine or modify meaning in a situation. Annells (1996) notes that these concepts are open to criticism and interpretation by scholars, calling social interactionism a “microsociological theory” (p. 381) because it focuses on the individual, not the larger social environment. However, despite criticisms, practitioners of grounded theory assume that reality is a social construct and that theory can be derived from the social processes individuals experience to make sense of a phenomenon.

Glaser and Strauss eventually disagreed about the direction grounded theory should take, and Strauss collaborated with Juliet Corbin to evolve the methodology (Strauss & Corbin, 1990). Traditionally, grounded theory reflects a positivist and objectivist worldview that assumes an external reality, although Strauss and Corbin moved grounded theory into a more postpositivist light by advocating for unbiased data collection, and a fair representation of the participants’ voices (Charmaz, 2003; Strauss & Corbin, 1990). Strauss and Corbin offered specific procedures for conducting a grounded theory study in *Basics of Qualitative Research* (1990), but Charmaz (2008) argues that this caused researchers to design their studies too concretely. Despite the evolving nature of grounded theory, Charmaz (2003) argues that all forms espouse strategies including simultaneous data collection and analysis, emergence of themes, discovery of basic social processes, category construction that explain these processes, theoretical sampling, and the integration of categories into a theoretical framework. Traditional grounded theory is paradigmatically aligned with postpositivism and based in critical realist and objectivist

philosophies. However, a version of grounded theory that emphasizes subjectivity and a relative perspective is moving the methodology into the constructivist paradigm (Annells, 1996; Charmaz, 2006).

2.4.1.2 Constructivist Grounded Theory

In response to her position as a social constructionist researcher, Charmaz adapted the methodology she named constructivist grounded theory (2006; 2008). In contrast to Glaser and Strauss who did not consider that the researcher might influence the analysis (Charmaz, 2008), researchers using constructivist grounded theory assume that there are multiple realities, and that the researcher and the research participants co-construct and analyze the data (Charmaz, 2003; 2006). Constructivist grounded theory argues that data does not reflect an objective reality; that is, the data offers a view of the participants' world that is influenced by time, by cultural and structural contexts, and by the researcher, the participants, and the research process (Charmaz, 2003; 2006; 2008). Therefore, constructivist grounded theory methods are tools for discovery, not conveyors of truth. Further, researchers also have preconceptions, experiences, and biases that may color the research experiences and must be handled reflexively. Charmaz (2006) suggests that the research question should shape the research methods, and she offers suggestions for conducting a grounded theory study including strategies for sampling, semi-structured interviews, data analysis, memoing, and for assessing the quality of a study.

2.4.2 Data Collection: Sampling

Initially, purposeful sampling is done to identify participants who may provide rich data about the research question (Charmaz, 2006). However, theoretical sampling is a core component of grounded theory that is useful once data is analyzed through constant comparative methods and themes and categories begin to emerge. Theoretical sampling is a strategy used to flesh out emerging categories and theory (Charmaz, 2006). In particular, a researcher may ask participants new questions, or follow-up with, or recruit new participants. Memo writing is a useful tool for determining the direction of

theoretical sampling. Theoretical sampling is particularly useful for identifying the characteristics of a category, identifying relationships between categories, to make the analysis more abstract, to ground the theory in the data, and to make analytic connections between categories (Charmaz, 2006).

Finally, theoretical sampling may lead to saturation of theoretical categories. Categories are considered saturated when no new insights are forthcoming, although Charmaz (2006) cautions that saturation is not the same as seeing similar phrases or events. Moreover, early saturation may result if the researcher does not critically analyze categories. To evaluate whether saturation has been reached it is important to have a good understanding of the comparisons of category properties and of the relationships between categories. It is then important to assess if and how these comparisons provide theoretical insight, specifically if new relationships or themes need further development. Saturation is achieved when no new patterns or abstract analyses between categories emerge (Charmaz, 2006).

2.4.3 Data Collection: Semi-Structured Interviews

Charmaz (2003) offers a guideline for conducting constructivist grounded theory interviews that focuses on locating a participant's story within a basic social process. According to Charmaz, interviews begin by defining a central problem (e.g., "Tell me about what happened when you were first diagnosed with *condition of interest*?") and evolve as the researcher and participant co-construct the data. Constructivists emphasize the importance of context, whether of the interview, the participant's experience, or the context of the research question within larger society (Charmaz, 2003). Since constructivist grounded theorists are attempting to uncover the social processes that shape events, interview questions should reflect symbolic interactionism, and "be sufficiently general to cover a wide range of experiences as well as narrow enough to elicit and explore the participant's specific experience" (Charmaz, 2003, p. 315). Charmaz suggests using a conversational interview style using open-ended questions. The interview script will likely evolve as participants are interviewed and new questions arise (Charmaz, 2003), thus multiple interviews with each participant may be necessary to

fully explore emerging themes and to ensure that data saturation is reached. This is especially important considering that multiple realities exist, and that perceptions and experiences may change over time. Charmaz (2003) also argues that multiple interviews serve as “independent checks” (p. 318) and allow the researcher to probe for more details that may enhance emerging theory.

2.4.4 Data Analysis

The process of data collection and data analysis is iterative and emergent, and grounded theory coding occurs in three phases including an initial phase in which the researcher codes a word, line, or segment of data, a focused phase that uses the most frequently occurring initial codes to sort and organize the data, and theoretical coding to analyze increasingly abstract ideas about the data (Charmaz, 2006). Charmaz (2006) argues that the researcher is not “neutral” (p. 46) when coding because he or she codes using language that is based on a particular worldview. Charmaz (2006) suggests using action codes during initial coding (namely gerunds), arguing that this prevents the researcher from developing theories too early. In addition, *in vivo* codes, codes using the symbolic language used by participants, may be useful for merging common phrases into the theory (Charmaz, 2006). Initial codes should be open-ended and “grounded” in the data. Importantly, the initial codes provide the researcher with the tools to understand where more information needs to be collected, to flesh out data, and to determine the “fit and relevance” of the data (Charmaz, 2006, p. 54). Initial coding may be conducted word-by-word, line-by-line, or incident-by-incident.

After a list of codes has been developed, and a number of transcripts have been reviewed, Charmaz (2006) suggests that the next step is focused coding. Focused coding is done to organize the data using the initial codes that occur most often, and it checks the relevance and analytic value of the initial codes (Charmaz, 2006). At this stage, some codes will be collapsed into themes. Constant comparative methods are an integral part of focused coding (Charmaz, 2006).

Constant comparative methods (Glaser & Strauss, 1967) are an important part of grounded theory analysis, and are used to collapse codes into more abstract categories.

Stanley (2006) describes that constant comparative analysis involves comparing all coded data within each category to move from simple description of categories to theory.

Constant comparative analysis in grounded theory includes comparisons of participants' responses, the data from the same participant at different time points, incidents, data with categories, and a comparison of categories with categories (Charmaz, 2003; Ghezeljeh & Emami, 2009). By using constant comparative methods, statements made in the same interview are compared, and then they are compared with statements in other interviews (Charmaz, 2006). Constant comparison is an on-going process that occurs throughout data collection and analysis.

The final step in grounded theory coding is theoretical coding. Theoretical coding involves assessing the relationships between data coded during focused coding (Charmaz, 2006). At this point, it is helpful to draw diagrams to help sort the data, and to make comparisons between categories to begin to develop theory. Glaser (1978) argued for the six C's to examine categories based on causes, consequences, contexts, contingencies, covariance, and conditions. Charmaz (2006) argues that using these categories can strengthen the analysis and the emerging theory. Theoretical coding both guides theoretical sampling and identifies relationships between categories (Stanley, 2006).

Theoretical sorting, diagramming, and integrating are important components of theory development in grounded theory (Charmaz, 2006). Theoretical sorting of memos encourages theory evolution by abstractly combining and comparing categories (Charmaz, 2006). Sorting and diagramming may be useful tools for organizing data and integrating categories (Charmaz, 2006).

2.4.5 Memoing

Memoing is a critical analytical and reflexive tool for grounded theorists, and Charmaz (2003) considers memoing the step that bridges coding with drafting the research report. The purpose of memoing is to record hypotheses and thoughts about emerging themes, and to reflect on the interaction between the researcher and the participants as themes evolve from the data (Mills, Bonner, & Francis, 2006). Memoing may also increase abstract thinking (Charmaz, 2006), and a memo provides a schematic for how thoughts,

data collection, codes, and categories evolve (Ghezeljeh & Emami, 2009). However, constructivist researchers should keep abstract thinking firmly grounded in the data that was co-created by the participant and the researcher (Mills, Bonner, & Francis, 2006). Importantly, memo writing is essential for capturing the researcher's biases and preconceptions, the interplay of researcher and participant, and other elements that influence the data (Mills, Bonner, & Francis, 2006).

2.4.6 Developing Theory

“Interpretive theory calls for the imaginative understanding of the studied phenomenon. This type of theory assumes emergent, multiple realities; indeterminacy; facts and values as inextricably linked; truth as provisional; and social life as processual” (Charmaz, 2006, p. 127). Theory is established by making connections between, and asking questions about, the data, which leads to abstract thinking (Charmaz, 2006). Using gerunds to code data, and studying processes are helpful tools for theory development. Charmaz (2006) cautions against coding for themes rather than actions which may lead to descriptions rather than theory. Again, memo writing is an integral part of theory development because it enables the reader to think carefully about the data and connect categories (Charmaz, 2006).

2.4.7 Assessing Qualitative Research: Trustworthiness

This section describes procedures that can enhance the rigor of qualitative research; specific strategies used to ensure the trustworthiness for each study will be described in Chapters III-VII and discussed in Chapter VIII. The constructivist paradigm acknowledges the importance of the researcher, including her/his opinions, beliefs, and perceptions, in co-constructing data (Morrow, 2005). Researcher bias is both acknowledged and embraced. Thus, reflexivity—the practice of engaging in an “explicit and self-aware meta-analysis of the research process” to evaluate if and how findings are influenced by “subjective and intersubjective elements” (Finlay, 2002, p. 531)—is an essential component of qualitative research that allows the researcher to examine her/his own biases, perceptions, theories, and worldview in order to understand how they help

shape the data. There is no consensus amongst qualitative researchers about how to best assess the rigor and quality of qualitative inquiry (Rolfe, 2006; Sandelowski & Barroso, 2002; Morse et al, 2002). One widely accepted approach is to use criteria including credibility, transferability, dependability and confirmability to evaluate the trustworthiness of qualitative research (Guba, 1981; Lincoln & Guba, 1985). This approach corresponds to criteria—including internal validity, external validity and generalizability, reliability and objectivity—that is commonly used to evaluate positivist or quantitative research (Guba, 1981; Shenton, 2004).

Lincoln & Guba (1985) consider credibility to be the primary and perhaps most significant criteria for evaluating the trustworthiness of qualitative inquiry. Credibility—the degree to which a “true picture” (Shenton, 2004, p. 63) of the research question and participants’ responses is portrayed—can be enhanced by a number of strategies including—but not limited to—triangulation, member checking, iterative data collection and analysis, frequent debriefing sessions with advisors, and peer review of the study findings (Shenton, 2004). Transferability reflects whether findings resonate with—or can be applied—to other settings. Researchers need to provide sufficient detail about the research setting and the participants in order for a reader to be able to decide whether findings are applicable elsewhere. Dependability—often difficult to achieve in qualitative work (Shenton, 2004) because findings are based on the interpretation and co-construction of data between researchers and participants—can be established by providing adequate details about study design and methods for another researcher to follow the data collection and analysis procedures and recreate the study. Finally, methods described previously—including triangulation and sufficient detail about research methods and setting—can help readers assess the confirmability of qualitative research. The researchers’ reflexivity and an audit trail of research procedures is important for establishing confirmability; that is, researchers must demonstrate that findings ‘emerged’ or were identified through rigorous data collection and analysis, and are therefore not the result of the researchers own biases and preconceived ideas (Shenton, 2004).

2.5 Summary

This chapter includes a description of the research setting and study participants, a summary of phenomenology, photovoice and grounded theory, and strategies for ensuring the trustworthiness of qualitative research. Proponents of all three methodologies state that the proposed methods are flexible, and that researchers do not have to adhere to them prescriptively. Instead, methods may be modified to fit the context and goals of the study, to encourage creativity, and to meet the needs of the participants (Charmaz, 2006; van Manen, 1990; Wang & Burris, 1994; 1997). A detailed description of the data collection and analysis procedures for each study will be described in the relevant chapters.

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Chapter 3

3 Hard to Swallow: A Phenomenological Exploration of the Experience of Caring for Individuals with Myotonic Dystrophy (DM1) and Dysphagia

3.1 Abstract

Background: Myotonic Dystrophy (DM1), a genetic, multi-system disorder, is the most prevalent adult form of muscular dystrophy. Dysphagia is a common symptom that may be difficult to diagnose and treat and can be associated with increased morbidity and mortality. Pre-existing cognitive impairment or apathy, both well described in the DM1 literature, may contribute to management challenges. Caregivers may become important for managing a family member's swallowing dysfunction. While clinicians place great importance on swallowing difficulties, it is unknown how dysphagia impacts patients and their caregivers. Therefore, the purpose of this study was to explore the experiences of caregivers living with those with DM1 and dysphagia.

Methods: An interpretive phenomenological approach was used to study the lived experience of six caregivers for individuals with DM1 and dysphagia. Audio-taped semi-structured interviews were used for data collection, and data was analyzed using van Manen's steps for phenomenological analysis.

Results: Despite the potential for dysphagia to cause morbidity and mortality in DM1 individuals, caregivers did not describe this as a problematic symptom. Instead, they described more debilitating symptoms like fatigue or weakness and discussed the caregiving experience. Themes pertaining to participants' *lived body*, *lived relationality*, *lived time* and *lived space* were identified.

Conclusion: Health care providers need to balance issues of clinical concern with those that are important for individuals and their family members. Assessments of caregiver knowledge and burden at each clinic visit may be warranted.

3.2 Introduction

Myotonic dystrophy (DM1) is an inherited, chronic, and progressive neuromuscular disorder that may occur rarely at birth (congenital form) or more commonly manifest during adulthood. Given the autosomal dominant inheritance, DM1 generally affects roughly 50% of every generation of a family. Complications of DM1 include muscle weakness, fatigue, hypersomnolence, cardiac conduction deficits, cognitive dysfunction, apathy, and gastrointestinal manifestations including dysphagia (Harper, 2001; Turner & Hilton-Jones, 2010).

Dysphagia is defined as difficulty swallowing foods or liquids safely that may result in malnourishment or an aspiration pneumonia that occurs when foreign substances enter the lungs (National Institute on Deafness and Other Communication Disorders [NIDCD], 2014). Swallowing difficulties are common in DM1, and may affect between 25% to 80% of patients (Bellini et al., 2006; Ronnblom & Danielsson, 2004). Individuals experiencing dysphagia may report coughing while eating, heartburn, chest pain, or reflux (Bellini et al., 2006). Dysphagia may cause embarrassment and lead to decreased social participation, low self-esteem, and poor quality of life. Relationships with caregivers may also be negatively affected by an individual's inability to share meals (Ekberg, Hamdy, Woisard, Wuttge-Hanning & Ortega, 2002; Plowman-Prine et al., 2009). Health care professionals consider dysphagia particularly serious in DM1 due to the risk of sudden death from choking (Garrett, DuBose, Jackson, & Norman, 1969). Moreover, in addition to respiratory failure and cardiac problems, aspiration pneumonia is a common cause of death for individuals with DM1 (de Die-Smulders et al., 1998; Mathieu et al., 1999; Turner & Hilton-Jones, 2010).

However, diagnosis of dysphagia in DM1 is complicated because patients rarely complain about or describe difficulties with their swallowing. Remarkably, patients with DM1 did not report symptoms of dysphagia despite evidence on videofluoroscopy suggesting abnormal swallowing function (Leonard, Kendall, Johnson, & McKenzie,

2001). It is possible that muscle weakness and swallowing dysfunction progress so slowly that individuals develop compensatory strategies that leave them unaware of their dysphagia (Leonard et al., 2001). Moreover, findings from a survey study of 360 frail elderly patients suggest that participants believed that dysphagia was untreatable, and therefore there was little to gain by mentioning symptoms to health care providers (Okberg et al., 2002). It is unknown if this phenomenon occurs in DM1

Apathy and cognitive impairment may further complicate the diagnosis and treatment of DM1-related dysphagia (LaDonna, Koopman & Venance, 2011). In particular, there is a well-described cognitive and personality profile for DM1 individuals that may be characterized by low IQ, an avoidant personality, lack of motivation and difficulty with executive function (Delaporte, 1998; Meola et al., 2003; Sistiaga, et al., 2010). Clinical experience suggests that some individuals with DM1 manifest apathy and therefore may minimize their symptoms, miss clinic appointments, and may not express interest in their health (Meola & Sansone, 2007). While speculative, it is possible that these characteristics impact an individual's ability and/or willingness to recognize and manage swallowing dysfunction.

As a result, informal caregivers may play an important role in assessing and managing swallowing dysfunction for their loved ones. To our knowledge, there are relatively few studies researching the experiences of caregivers for adults with muscular dystrophy (Boström & Ahlström, 2005a, 2005b; Boström, Ahlström, & Sunvisson, 2006; Boyer, Drame, Morrone, & Novella, 2006; Cup et al., 2011; Timman, Tibben, & Wintzen, 2010). These studies explored a variety of muscular dystrophies using quantitative and/or qualitative methods, and did not specifically address the implications of caring for individuals with DM1 and swallowing dysfunction. However, a qualitative study explored nine individuals who cared for loved ones experiencing dysphagia after stroke or other traumatic injuries and found that caregivers prompted their family members about safe eating practices, spent extra time preparing meals, and experienced a sense of fear or worry (Johansson & Johansson, 2009).

While clinicians provide surveillance for swallowing problems and are concerned about the potential for dysphagia to increase the risk of morbidity or mortality, it is unknown if and how swallowing problems impact the daily lives of DM1 patients and their identified caregivers. Therefore, it may be important to consider the potential disconnect between health care provider's clinical concerns and patients' and family members' perceptions. However, much of the DM1 literature considers dysphagia largely from the clinicians' perspectives, therefore little is known about the experience from the perspectives of the patients and caregivers. Therefore, the purpose of this research is to explore the experiences of providing care for a person with DM1 and known dysphagia. Findings may influence clinicians' approach to dysphagia diagnosis and treatment; that is, findings may help identify appropriate questions to ask patients and caregivers during clinical encounters, and/or to refine management strategies.

3.3 Methodology

The methodological framework used to guide this study involved a philosophical inquiry of an interpretive phenomenological nature as articulated by Merleau-Ponty (1962) and van Manen (1998). Using the viewpoints of van Manen (1990) and Merleau-Ponty (1962), we explored each participant's experience of embodiment, as being situated and relational, or as "being-to-the-world". This refers to the idea that human knowledge is relational, temporal, and present in the world, instead of just being a static object in the world, independent of the researcher. According to van Manen (1990), Merleau-Ponty offers four existentials: "1. Lived body (Corporeality) 2. Lived space (Spatiality), 3. Lived time (Temporality), and 4. Lived human relation (Relationality)" (p.101). These four existentials were utilized and reflected upon throughout the research process to understand the realities of the participants and their lived experiences and to allow the findings to be grounded on the fundamental life world structures which all human beings experience, although not all in the same ways. The four existentials of lived body, lived space, lived time, and lived human relation are categories for phenomenological questioning, reflecting, and writing (van Manen, 1990).

3.3.1 Methods

For the purposes of this study, a caregiver was defined as a spouse, family member or friend who provided some degree of supportive care to a person living with DM1. We designed the study and collected and analyzed the data using an interpretive phenomenological approach (van Manen, 1990). Interpretive phenomenology seeks to explore and understand the nuances of lived experience by uncovering taken-for-granted assumptions (van Manen, 1990). Individuals with mild to moderate DM1 and swallowing problems (by physician report) who attended an outpatient neuromuscular clinic were invited to identify and approach a caregiver about the study; six caregivers (two females; four males) agreed to participate. Data was collected during semi-structured interviews lasting up to an hour; the interviews began by asking participants to reflect on the meaning of the term ‘caregiver’. Next, the interviewer (KAL) used a series of prompts to probe about awareness and understanding of swallowing dysfunction and other DM1-related symptoms, strategies for management, and symptom impact on daily life. All interviews were recorded and transcribed verbatim.

Data was analyzed using van Manen’s (1990) suggested steps for phenomenological analysis. Two researchers (KAL and WJK) began by reading each transcript twice to gain a holistic sense of the data, and then independently chose a sentence or phrase that captured the essence of each transcript. ‘Caregiving is a full-time job’ and ‘caregiving is a partnership’ are examples of sentences that described the essence of two transcripts. Next, a selective reading approach was independently used to code the data and extract statements that described the caregiving experience. KAL and WJK independently underlined key passages from the text related to dysphagia or caregiving and developed a list of codes using words or phrases that reflected the participants’ experiences. KAL and WJK then met to discuss their codes and combined them into overarching preliminary themes. Key quotes supporting the themes were extracted from the data. The themes and quotations were then discussed with all authors, and a final list of findings was developed by consensus. Recruitment ceased when no further themes emerged. This does not mean, however, that new information would not have been gained by further data collection; rather, the data collected was sufficient to allow a rich understanding of the

phenomenon under study. As suggested by van Manen (1998) the use of temporal, spatial, relational, and bodily existentials to guide the analysis yields a richly-textured understanding of the embodied nature of caregiving for those with DM1 and dysphagia.

To further ensure the rigor of our study (Morse, Barrett, Mayan, Olson, & Spiers, 2002), we sent a letter to participants that described the preliminary findings, and invited each caregiver to participate in a second interview to comment on the emerging themes. Only two participants agreed to participate in follow-up interviews.

The Western University Research Ethics Board approved this study. To protect the confidentiality of our research participants, we have assigned each person a pseudonym. At the time of his follow-up interview, Sam was a 79-year-old living with his moderately affected wife. Sam has three children with DM1 and a grandchild with congenital DM1; Sam also provides some degree of care for his moderately affected son. Laura is in her 60s and lives with her mildly affected husband. Laura also has a son with moderate DM1 for whom she does not consider herself a caregiver, although she often runs errands for him. Daisy is a caregiver for her moderately affected husband, and she also works outside of the home. Michael is a retired gentleman in his 60s who lives with his mildly affected wife, and John is in his 50s and he cares for his mildly affected wife. Finally, Greg is in his late 40s and provides care for his mildly affected partner and for her son with congenital DM1. Greg's affected father-in-law died unexpectedly following a choking episode.

3.4 Results

While dysphagia is one of the most life-threatening symptoms of DM1 (Harper, 2001; de Die-Smulders, 1998), five of the caregivers in the present study did not consider it to be their family members' most problematic symptom. Instead, most identified fatigue and weakness as symptoms that impacted their affected family members' function and quality of life. However, despite the interviewer's attempts to circle the conversation back to the experience of caring for someone with dysphagia, participants chose to focus on their role as caregivers rather than on their loved ones' symptoms. While caregivers offered insight

into their beliefs about DM1 and swallowing dysfunction, and were able to describe strategies for managing symptoms, the participants continually returned to discuss their caregiving roles and responsibilities. This divergence from the original research question is not wholly unexpected given that the purpose of interpretive phenomenology is to have participants reflect on their lived experience and to uncover taken-for-granted assumptions (van Manen, 1990). We identified five themes that reflected the bodily, spatial, temporal and relational existentials described by van Manen (1990):

Relationships Drive Perceptions; The Meaning of Caregiving; The Physical Experience of Caregiving; Carving Out a Space for Respite, and Looking Into a Crystal Ball.

3.4.1 Relationality—Relationships Drive Perceptions

Relationality refers to the lived relations and spaces that are shared with others (van Manen, 1990). In particular, sharing human relationships with others develops impressions of others that are confirmed—or not—through interactions. For instance, participants' relationships with their affected family members influenced their beliefs about the meaning of caregiving and the impact of DM1 symptoms on their lives. For example, it was evident that health care providers were concerned about the swallowing function of several DM1 patients since Sam, Daisy and Michael all stated that their family member had been referred for a videofluoroscopic swallowing assessment, yet participants were largely unconcerned about their swallowing dysfunction. This was despite being shown video evidence of their loved ones' swallowing abnormalities. Sam described that it was “the most fascinating thing to see this food go back, stop, and then go down”, yet he did not consider swallowing to be a particularly troubling symptom because his day-to-day experience of living with his wife reinforced his belief that she had developed appropriate compensatory strategies. However, a health care provider would likely be concerned about the potentially serious strategies he described:

So, as far as that's concerned, she has a way...we have morning vitamin pills. There was about 6 or 7 when washed down and they go down then the Omega 3, 6, 9, they're a lot bigger so but she has an excellent way of popping them back up. So in that sense she doesn't have a problem swallowing. But like yesterday, we

had stew so she just put it through the blender. Because I think more than just swallowing problem it is her muscles are weak—too weak—and so she says that's enough...and that's obviously related to the myotonic. But, so in a way, swallowing she handles quite well.

Moreover, Greg's father-in-law died following a choking episode, an occurrence that made his partner more vigilant about her swallowing function. However, Greg was less concerned about swallowing abnormalities; instead, he focused on his partner's difficulty breathing.

My concern is the breathing more so than her choking. Everybody can get out of a choking situation, but the breathing problem is the big issue. She keeps losing percentages of her lungs' capacity because of her diaphragm. That's my big concern.

This was surprising given his father-in-law's unexpected and sudden death from choking and his partner's concern about her swallowing dysfunction. Further, respiratory failure is a slowly progressive symptom as opposed to choking which can cause an acute complication like aspiration pneumonia or sudden death. However, Greg seemed confident that he could help his partner should she choke, yet seemed to feel helpless about the progressive decline of her lung capacity. The fear of losing his partner to a symptom or event that he could not control may have influenced how he evaluated the impact of his partner's symptoms, and in turn, where he chose to focus his energy.

In general, dysphagia appeared to be a symptom that participants felt the individual with myotonic dystrophy had compensated for and managed; therefore swallowing dysfunction did not seem to cause great concern or to affect shared experiences like mealtimes or social activities. In particular, caregivers described that their loved ones were aware of their limitations, and had identified strategies like avoiding certain foods, cutting things into smaller pieces, or having liquids with meals to facilitate safer swallowing.

As she told the fellow we were talking to, she's very careful how she eats. And

by careful, I mean she cuts small pieces. She eats very slow. I could probably eat two meals to her one (Michael).

Instead, caregivers considered weakness and fatigue far more troubling symptoms. Fatigue impacted the social participation of caregivers and their affected family members, and seemed to negatively affect the marital relationship. Daisy described that it was not uncommon for her husband to fall asleep at inappropriate times, and Greg's partner stated that Greg must feel like he "lives alone" because she falls asleep so frequently. Moreover, Sam noted that his wife's fatigue, coupled with her multitude of complications, made it difficult for her to be motivated to be active or social. He regularly had to prompt her to leave the house to "get some fresh air". Laura and Michael described changing their expectations about, and opportunities for, social outlets. Michael described staying in to play cards instead of engaging in activities outside of the home, and Laura prepared special meals to ensure that her husband could participate in meals with friends:

We know they (*husband and son*) have muscular dystrophy and we accommodate time or meals so that...basically, if we have company and I'll make sure it it's something that maybe not 100% of the meal (*husband*) can eat but a good portion—75% maybe even 80%-- and he wants that too because then it's – he's not depriving me of doing things.

3.4.2 Relationality— The Meaning of Caregiving

While participants did not speak at length about assisting their affected family member with managing swallowing dysfunction, they did speak in-depth about the overall caregiving experience. The data suggested that there were three main reasons for being a caregiver: (1) It is the nature or personality of the individual to be a carer; (2) There is no one else to do the caregiving, and (3) Caregiving is done out of love. Five of six participants endorsed that 'caregiver' was a term that they would use to describe themselves. Sam put caregiving for his wife in context with his volunteer work as a palliative caregiver in a hospice:

Well, it's interesting. Because in a way, it's different than with the palliative care. I always felt that we gained an awful lot by giving time to chat and so on. It was very enriching in many cases. With my own wife, it's not so much enriching as to feel that I have made it a little more comfortable and I have been able to assist her in something that she found difficult or because we've been married for 48 years...so we've exchanged a lot... And, it isn't quite the same.

On the other hand, Laura felt that she was not a caregiver because her spouse and child “manage on their own”. For Laura, a ‘caregiver’ is a defined role and title that must be earned by performing specific tasks. She viewed her work checking on her son and preparing meals for her husband not as caregiving duties, but as intrinsic parts of her lived relations as a wife and mother

Um...well, the word caregiver is used and it's used for people who actually need that kind of care and I think – I guess mostly personal care, feeding and all of that. So, I'm not a caregiver for either one of them. But I help when I can.

Similarly, for Sam and Michael, caregiving was seen as part of the marital relationship that would be present even without factoring in a chronic disease.

Well, I guess I could look at it that way too. ... definitely [she] does what she's capable of doing. And I guess I do that for her too. In my opinion, she would be a caregiver of me too. To me it's a 50/50 deal. You don't know what life's going to deal you. You've got to make the best of what you have and go from there.

(Michael)

Also, some participants seemed to have a nurturing nature in all facets of life that influenced their experience of caregiving. For Sam, his experience of caregiving transcended caring for his family members with a chronic, progressive disease. Sam's caregiving took many forms throughout his life, both as an educator and as a volunteer in a hospice. He perhaps chose these vocations because of an inherent need to serve and support; in turn, he used these experiences to shape how he cared for his wife.

I was a teacher for 31 years and there definitely is caring for the kids that they

succeed. Now with [spouse] it definitely is love, I mean she is my wife. And it has become, or it has become a little bit more. Just last night she said, how did she put it, you are doing things that I used to do...

In contrast, John's experience of caregiving was one of burden and burnout. John described being responsible for all of the household chores in addition to working full time, and he resented what he perceived as his wife's learned helplessness: "She needs to get back on her CPAP and it's just finding the phone number for her. And she expects me to do that but that's not my job. 'I'm not doing that for you dear. That's drawing the line there, you can call.'" When asked if there were any rewards of caregiving, he noted that the one positive aspect of caregiving was the knowledge that his wife was receiving appropriate care:

No. Other than I know she's getting good care. So, you know, that's the biggest thing. She's being taken care of. 'Cause I know what else is out there...But, no, other than I know she's getting good care and being taken care of. Not being abused by anybody or nothing, so...Yeah, that's about it. (John)

3.4.3 Corporeality—The Physical Experience of Caregiving

"Lived body" (corporeality) refers to the phenomenological fact that we are always bodily in the world" (van Manen, 1990). For most participants, their physical body affected either how they interpreted their loved one's symptoms or how they were able to provide care. Despite not being cognizant of the extent of their loved one's swallowing dysfunction, some caregivers had good awareness of their loved one's bodies. Caregivers knowledgeably discussed the main DM1 symptoms and the variability of disease progression. Caregivers were readily able to describe the "lived body" of fatigue, cardiac and respiratory problems, dysphagia, and weakness as primary symptoms affecting their family members. For example, Michael was mindful about taking objects from his wife because her myotonia made it difficult for her to release her grip:

Well, probably lots of times I help her get dressed, maybe do up her shoes for her. The big thing is getting stuff out of the cupboards because of with her grip. I'm

going to have to do that type of thing. Or you've got to be careful when ... if she hands me something I've got to realize that it takes her a while to let go of it, so if I don't, like sometimes pull her over or pull her arm type of thing.

Participants described that the experience of caregiving involved both physical and emotional work. Most caregivers cared not only for their family member with DM1, but for other affected family members and/or aging parents. Participants described that the work of caregiving involved the bodily work of cooking, cleaning, heavy-lifting, running errands, and assisting with bathing and dressing. Participants also discussed that attending and asking questions at medical appointments were important aspects of their roles that facilitated the work of caregiving. The task of 'prompting', whether to take medication, chew slowly, or for their family member to be social, was described. Moreover, Sam used his body to provide comfort and healing for his wife: "And I do Reiki, I give her Reiki treatment. We do massage, light massage with maybe a bit of aromatic oil." However, caregivers' physical limitations sometimes impacted their ability to perform the physical tasks of caregiving. John described having a pinched nerve, and Sam stated that a shoulder injury affected his ability to assist his wife in lifting or carrying heavy objects.

Caregiving also enacted an emotional toll on Greg and John who described that the constant duties of caregiving were "annoying" and frustrating. In particular, John stated that he felt "down", and his emotions affected him both physically and emotionally to the point that he was on a number of prescription medications to treat his anxiety and depression. Overall, caregivers described that much of the perceived effort of caregiving was found to be emotional, with "worry" as a prominent theme. Participants disclosed a variety of concerns, including whether their family member was eating enough, the impact of DM1 on their own physical and emotional health, increased caregiving tasks as DM1 progresses, and the constant fear of the likelihood of an early or unexpected death.

You know, that's the most important right there cause you never know one day he could be lying dead somewhere and I'd never know it.... Like I say, you never know if he could end up one day in bed dead, you know, and I wouldn't know it....

That's something I don't want to think about. You know, nobody wants to think about that but if it happens it happens. (Daisy)

3.4.4 Spatiality—Carving Out a Space for Respite

Spatiality, or lived space, is “felt space” that describes how individuals experience spatial dimensions in their everyday lives (van Manen, 1990). In general, participants described that it was difficult to find physical spaces that accommodated individuals with a disability, and navigating in public spaces was impacted by their loved ones’ bulky mobility aids like wheelchairs or oxygen tanks. As a result, social circles and outlets diminished.

Other than going places – that’s the biggest drawback, going and figuring out whether we need to take her machine and all the stuff, and when it’s really hot out she doesn’t bother going outside, it’s so humid. (John)

Moreover, the emotional toll that Greg and John described also affected their ability to feel comfortable and relaxed in their homes because they always felt that they should be performing tasks or supporting their partners. Therefore, for John and Greg, caregiving constrained their ability to achieve a physical distance from their duties and responsibilities.

They’re (*caregiving duties*) intense right now as far as I’m concerned because I can’t go out on my own without worrying about her. As I said, we travel with cell phones, so if I’m out and about, I’m always close. I can’t go anywhere without always worrying about her. So I don’t go too far away. I stay close to the house. So it’s really restricted my life in that respect. I don’t want to go out and get a part-time job, because if I do that, I’m on the job, she phones you up and she has trouble, what am I going to do? (Greg)

Therefore, there was a sense that burden was part of the experience of caregiving for John and Greg. To cope with the stress of caregiving, they attempted to carve out both an emotional and a physical lived space for respite.

She just takes it (*caregiving*) for granted. Which can be real annoying. I try to sit down and relax and she's going jibber, jibber at me. Shhh, I'm trying to relax. So I just end up going sitting on the porch or going out in the back yard with the dogs. Here we go, here's some relaxing (John).

3.4.5 Temporality—Looking Into a Crystal Ball

Temporality refers to subjective time rather than 'clock time', and an individual's 'temporal landscape' is colored by the past, present and future (van Manen, 1990). Prior to their loved one's diagnosis, few participants had knowledge about DM: "Like I've watched the Jerry Lewis Telethon and there's a bunch of families there with different muscular dystrophy diseases but coming up with myotonic dystrophy—no. No, I [had] never heard of the word before (Daisy)." Daisy also reflected on the changes in their lives since her husband's diagnosis; they had previously enjoyed parties and being social, but now spent more time at home alone. Further, Sam discussed the past with a sense of regret and wondered whether he and his wife would have had children had they known about the genetic nature of DM1: "We haven't dwelt on that. Once in a while it came up and you know what 'That's it...that's the way it is.'"

However, most participants focused on the present and living in the moment with their loved one. In particular, participants described living with their loved one's symptoms and adjusting to them as they progressed. Therefore, swallowing was not considered problematic because the affected individual was coping and managing in the here and now. Overall, there was a sense that while the complications of DM1 (namely fatigue and weakness) affected daily life, DM1 was manageable. Therefore, while some caregivers described a sense of regret or fear of the unknown about DM1, others like Laura, described DM1 as "no big deal"

We work with him rather than it dictates what we do. It's just--you just do it... there's no hardship at all. (Laura)

There was, however, greater concern for other symptoms including breathing difficulties and progressive weakness. Many participants had a 'crystal ball' mentality in which they

envisioned that their partners' decline in time was inevitable and that they would have to take on an increased caregiving role. Caregiving was already viewed as time lived as a consuming "24/7 job" (Greg) that was done in addition to full-time employment:

Like it's hard for me because I work and coming home, you know making meals and stuff, keeping our place clean and whatever, you know it's really hard, really really hard. (Daisy)

However, Michael, Greg and John discussed that their retirement afforded them more time as lived to devote to caregiving; this was positive for Michael and Greg, but not for John who was overwhelmed at the thought of being a full-time caregiver.

The experience of caregiving also involved future temporality as making plans and being prepared for eventual complications, and participants discussed their varying degrees of preparedness for the future. Only two participants (Sam and Greg) had previous experience with CPR or the Heimlich maneuver; however, they took first aid courses for employment or other purposes, not in preparedness for their loved one's impaired swallowing function. When prompted, however, all caregivers expressed an interest in learning more about what might be done in an emergency or unforeseen situation, but no one had plans to be formally trained. Despite this, most caregivers believed that they would be able to appropriately respond should a choking episode occur in future time.

Well if she were to choke, I could do the Heimlich on her. I plan on getting my CPR certificate. I'm working on that in (*location*). I'm waiting for a clinic to open up in case I ever need it. Then I'm certified. I used to be a trainer for a boy's hockey team. I had to get certified for that. So general first aid and stuff like that, I'm familiar with. I'm not totally lost when it comes to something like that. But CPR is what I've got to follow through on.

While there was a sense that caregivers understood that their loved one's condition would decline, the participants continually circled back to focus on the present time during the interviews. Overall, the experience of caregiving seemed to be one of love and duty,

tempered by the expectation that caregiving responsibilities would intensify in future lived time. The experience of caregiving for an individual with DM1, therefore, was influenced by their corporeality or lived body in lived time, lived space and lived relations when being with their affected family members.

3.5 Discussion

Given the attention, concern and surveillance that health care providers impart on DM1 affected individuals with dysphagia, we set out to explore caregivers' perceptions about, and their experiences with, their family members' swallowing dysfunction. However, the goal of interpretive phenomenology is to uncover taken-for-granted assumptions (van Manen, 1990), and study findings suggest that the overall caregiving experience, coupled with troubling symptoms like weakness and fatigue, have a far greater impact on the participants' lived experiences than dysphagia. Therefore, the study evolved into an in-depth examination of participants' knowledge and attitudes about DM1 and the meaning of their caregiving experiences. This speaks to the uncertain, fluid and emergent nature of qualitative inquiry (Lincoln, 1995; Whittemore, Chase & Mandle, 2001).

The participants in our study described their beliefs about DM1 and dysphagia, the impact of DM1 symptoms on their lived body, lived time, lived space and lived relations; in turn, the four existentials described by van Manen (1990) also influenced caregivers' descriptions of the meaning and work of caregiving. The relative lack of concern about dysphagia echoed other studies suggesting that individuals with DM1 are often not aware or concerned about swallowing or other consequences of their condition (Boström & Ahlström, 2005a; Meola & Sansone, 2007) despite significant concern from health care providers. This disconnect has been explored in other neurological disease populations, but to our knowledge it has not been studied in DM1. In a study of the multiple sclerosis population, for example, physicians tended to focus on mobility issues, while patients were more concerned about cognitive decline (Heesen, Kopke, Richter, & Kasper, 2007). In Parkinson's disease, while clinicians may attend preferentially to the motor features of the disease, it is often the non-motor features including mood problems, nocturia and drooling that most impact patients' health related quality of life (Martinez-Martin et al,

2011). Similarly, there is evidence that the cognitive and psychiatric features of Huntington's disease (HD) are far more disabling than chorea, yet research is often geared toward the motor symptoms (Bonelli & Hofmann, 2004). Finally, clinicians and patients may differ in their perception of the severity of HD symptoms, with clinicians objectively assessing symptom impact on disability, while patients subjectively assess symptom impact on daily life. This suggests that quality of life measures may be more indicative of symptom impact than disability scores (Banaszkiewicz et al, 2012).

Arguably, patients' description of symptoms may impact their caregivers' beliefs about how they are living with and managing their condition. In turn, clinical management may be influenced by patients' and caregivers' subjective reports of symptom impact on functional ability (Shulman, et al., 2006). However, caregivers were aware of health care professionals' concerns that dysphagia increases individual risk of morbidity and mortality related to choking, aspiration and sudden death. Others have hypothesized that this dichotomy may occur because there may be a distinction between knowing the symptoms of DM1 and fully understanding their implications (Boström & Ahlström, 2005a). We speculate that caregivers' lack of concern mirrors that of patients because the swallowing dysfunction evolves gradually over time allowing for strategies to compensate. A survey of 286 older adults with osteoporosis or osteoarthritis found that participants used a variety of strategies to compensate for their declining function including asking for help, giving up certain activities, performing behaviors that optimize their current function, and using assistive devices (Gignac, Cott & Badley, 2000). While this study did not speak to dysphagia, it does illustrate how individuals with chronic disease compensate for progressive functional loss. Our findings suggest that individuals with DM1 compensate for their abnormal swallowing by avoiding certain foods, having liquids with meals, or regurgitating objects that were not safely swallowed. For caregivers, therefore, there is no additional perception that swallowing is difficult to manage or is a hardship because their lived relations with their partners reinforce the belief that their loved ones have compensated appropriately.

However, similar to the experiences of thirty muscular dystrophy (n=10 individuals with DM1) caregivers described in a questionnaire study by Boyer, Drame, Morrone and

Novella (2006), the participants in our study expressed a sense of emotional and physical burden, primarily related to the anxiety of disease progression and sudden death. Further in line with our findings regarding the “work” of caregiving, five partners of DM1 individuals described that they increasingly became responsible for prompting their family members, for doing household chores, and for planning social activities (Cup et al., 2011).

Many of our findings resonate with the experiences of thirty-six caregivers for individuals with muscular dystrophy (19 DM1 caregivers were included) ascertained through inductive content analysis (Boström, Ahlström and Sunvisson, 2006). In particular, the sense that caregiving is shared with the individual living with muscular dystrophy and is done out of love or obligation were similar to our findings. Also, worry about disease progression and an increased care load, and the idea that individuals and families living with other conditions or life situations are worse off, resonated with our participants’ experiences. Using a hermeneutic qualitative approach, Cup et al (2011) found that the marital relationship can be affected by DM1 but that the five couples studied found ways to cope including giving each other space, respecting each other’s abilities, and allowing each partner to pursue his or her own interests. Moreover, we found that individuals with DM1 and their caregivers who see caregiving as part of the marital relationship and/or those who provide care out of love tend to view caregiving more positively. This is similar to studies that suggest that being in a healthy relationship was associated with the psychological well-being and better quality of life for individuals with muscular dystrophy and their partners (Boström & Ahlström, 2005b; Timman et al., 2010).

This is a small study reporting the experiences of six caregivers of individuals with DM1 and dysphagia and provides preliminary data that may inform future research questions. A longitudinal study with a larger sample that explores the experiences and processes of caregiving for individuals with DM1 and dysphagia may be warranted. Moreover, an exploration of patients’ experience of dysphagia, or an observational study exploring DM1 individuals’ daily activities, food choices and eating patterns may be helpful to guide the development of educational initiatives or interventions. In particular, future

research might consider appropriate questions to ask during clinical encounters or an examination of patients' condition-specific knowledge and their uptake of clinical information. Further, areas of education for health care providers might attempt to bridge the chasm between health care providers areas of concern with the complications that are most impactful to patients and their family members. Finally, a condition-specific assessment tool measuring swallowing function for individuals with DM1 may be useful (LaDonna, Koopman & Venance, 2011) to quickly assess for dysphagia during follow-up visits. While often outside a routine clinic visit, an assessment of caregiver burden for those caring for family members with DM1 may be necessary (Boyer et al., 2006). Health care providers may need to pay particular attention to the effect of DM1 on daily life for individuals and their families (Cup et al., 2011). While our results may not be immediately generalizable to the broader DM1 population, the findings may resonate with caregivers and other friends and family of those living with DM1.

3.6 Conclusion

In conclusion, while we set out to explore the experiences of caregivers for individuals with DM1 and dysphagia, we found that caregivers did not consider dysphagia a major concern. Instead they focused on other troubling symptoms like weakness or fatigue, and the impact of caregiving on their lived body in lived time, space and relations. This raises questions about the dichotomy between the concerns of health care providers and patients and their families, and how clinicians can assess the needs important to families while addressing clinical concerns. We suggest that clinicians emphasize the importance of monitoring and managing potentially life-limiting symptoms like dysphagia while also assessing how DM1 symptoms impact the quality of life for patients and their caregivers.

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Chapter 4

4 Picturing the Experience of Living with Myotonic Dystrophy (DM1): A Qualitative Exploration Using Photovoice

4.1 Abstract

Background: Myotonic dystrophy (DM1) presents with multi-systemic complications; moreover, there is a well-recognized DM1 personality profile that is characterized by executive dysfunction, an avoidant personality and impaired cognition. Understanding symptom impact on patients' lives is crucial for providing appropriate patient-centered care; however, much of the DM1 literature reflects the biomedical model and there is a paucity of articles exploring patient experience.

Objective: To use a novel research approach to explore DM1 patients' experiences.

Methods: Nine individuals participated in a qualitative study using the photovoice methodology. Photovoice uses the visual image to document participants' lives, and participants took pictures pertaining to living with DM1 that stimulated individual and focus group interviews. We used content analysis to analyze the data; in turn, codes were collapsed into themes and categories. Findings were presented to participants to ensure resonance.

Results: Participants took 0-40 photographs that depicted barriers and facilitators to living successfully with DM1. We identified two categories that include participants' challenges with everyday activities, their worries about the future, grief for lost function and social opportunities, and their resilience and coping strategies. Participants also described their experiences using the photovoice method.

Conclusion: Photovoice is a useful approach for conducting research in DM1.

Participants were active research collaborators despite perceptions that DM1-affected individuals are apathetic. Our findings suggest that participants' are concerned about symptom impact on reduced quality of life, not symptoms that clinicians preferentially

monitor. Nurses, therefore, are essential for providing patient-centered, holistic care for DM1 patients' complex biopsychosocial needs. Research exploring current physician-led clinical care models is warranted.

4.2 Introduction

Myotonic dystrophy (DM1)—the most common adult form of muscular dystrophy—is a chronic, progressive and inherited neuromuscular condition presenting with multi-system complications including distal muscle weakness, myotonia, hypersomnolence, early-onset cataracts, cardiac conduction abnormalities, and slurred speech and swallowing problems (Harper, 2001). DM1 patients' complex needs—including their cognitive and behavioral impairments—may complicate patient-centered care provision. In particular, patients' lower educational attainment, problems with executive function, and avoidant personality traits may make it difficult for them to take risks, make friends, or participate in new activities (Delaporte, 1998; Gagnon, Mathieu, & Noreau, 2007; Meola et al., 2003; Sistiaga et al., 2010); consequently, individuals with DM1 may be apathetic and disinterested in their health (Meola & Sansone, 2007).

Despite these challenges, it is essential that DM1 patients' values and experiences drive clinical care. Clinicians are concerned about the potential for cardiac, respiratory or swallowing abnormalities to cause morbidity or sudden death, yet research exploring patients' perspectives suggests that weakness, fatigue and myotonia have a greater impact on patients' quality of life (Boström & Ahlström, 2004; Cup et al, 2011; Gagnon, Mathieu & Noreau, 2007; Heatwole et al, 2012; Nätterlund et al., 2001). In particular, these symptoms often challenge DM1-affected individuals' ability to socialize, complete household chores, or engage in employment or educational opportunities. Moreover, progressive physical symptoms—coupled with patients' difficulties with motivation and planning—may impact patients' personal relationships (Cup et al, 2011). Consequently, patients may experience significant disruption and decreased satisfaction with employment and social recreation (Gagnon, Mathieu, & Noreau, 2007); in turn, impaired mental and physical function and reduced quality of life may result (Laberge et al., 2013).

However, despite evidence that clinicians and patients have different concerns and goals, much of the current DM1 literature is written from a biomedical perspective, and relatively few qualitative studies explore the perspectives of patients with muscular dystrophy (Authors, 2014; Boström & Ahlström, 2005; Boström & Ahlström, 2004; Boström, Ahlström, & Sunvisson, 2006; Cup et al., 2011; Faulkner & Kingston, 1998; Heatwole et al, 2012; Nätterlund, Sjöden, & Ahlström, 2001). These studies use a range of methods including content analysis and phenomenology to explore questions pertaining to living with muscular dystrophy; few of these studies represent the perspectives of North American patients, and individuals with DM1 are a small proportion of the overall sample. We believe that individuals living with DM1 have valuable insights into their condition and life experiences that can inform their health management, and that their voices should be more widely distributed in the literature. To address this, we used an innovative qualitative research method called photovoice (Wang & Burris, 1994; 1997) to explore the experience of living with DM1.

Photovoice is rooted in participatory action research (PAR) and was developed to study populations with low literacy residing outside of the traditional power structure (Wang & Burris, 1994). “Participatory Action Research (PAR) differs from most other approaches to public health research because it is based on reflection, data collection, and action that aims to improve health and reduce health inequities through involving the people who, in turn, take actions to improve their own health” (Baum, MacDougall, & Smith, 2006). While we do not believe that individuals with DM1 are ‘powerless’, we argue that their unique physical and emotional challenges, coupled with the power imbalance inherent in some medical encounters (Goodyear-Smith & Buetow, 2001) warrants a strategy that utilizes their strengths and experiences. In particular, photovoice uses photography to augment traditional interviews by offering participants a creative way to communicate their expertise, experiences, knowledge, and needs. In an effort to educate or enact change, participants may choose to disseminate their photographs and experiences to ‘stakeholders’ like clinicians or patient advocacy groups (Wang & Burris, 1994, 1997). Photovoice has been used with individuals with intellectual impairment (Jurkowski & Paul-Ward, 2007), acquired brain injury (Lorenz & Kolb, 2009), stroke (Levin et al., 2007), dementia (Genoe & Dupuis, 2013) and Alzheimer’s disease (Wiersma, 2011).

The purpose of this study was to add DM1 patients' voices to the literature; therefore, we used an innovative qualitative research approach to explore patients' experiences of living with a chronic and progressive neurological disease. In particular, we asked participants to reflect on the impact of DM1 symptoms on their daily lives, and to consider the barriers and facilitators to living successfully with DM1. We also sought to engage individuals with DM1 as research collaborators; the literature describes that DM1 patients may be apathetic or cognitively impaired, so we also asked the question: is photovoice an effective method for exploring the perspectives of individuals living with DM1?

4.3 Methods

Participants with mild to moderate DM1 (by physician report) attending an academic neuromuscular clinic were invited to participate. We purposively selected individuals who were able to provide informed consent and comply with study procedures; nine participants (four females) consented, and each was given a pseudonym to ensure confidentiality (Table 1).

We used a typical photovoice study consisting of a camera orientation session, an individual interview, and a focus group (Wang & Burris, 1997). Participants attended a camera orientation session in which they (1) discussed the ethics of picture taking, (2) were given a digital camera and instructed about its use, and (3) were asked to "take pictures of what it is like to live with DM1." Additionally, participants were asked to take pictures of people or things that either hindered or facilitated living successfully with DM1. Instructions were purposely kept vague to avoid the potential for researchers to influence picture taking. Participants were instructed that they had to seek written consent from each person they wished to photograph. Researchers asked questions to check the participants' comprehension and understanding of the task and instructions, and participants were given the opportunity to practice taking pictures.

Following the camera orientation session, participants had 2-3 weeks to take pictures and then returned for an individual interview. Participants were asked to describe each of

their photographs; in turn, their narratives directed the content and flow of the interviews. To further probe participants' responses, the researchers prepared general questions about their DM1-related medical history, their symptoms, and the impact of DM1 on their daily activities. We also asked participants to share their opinions about study participation and the photovoice method. Participants were then invited to choose 2-3 photographs to share during a focus group.

All 9 participants completed the camera orientation session and an individual interview. The first 7 participants were scheduled to participate in a focus group, and two focus groups were held with two and three participants each. Two individuals declined participation; one participant felt her fatigue precluded focus group participation, and one participant was lost to follow-up. All data was retained and analyzed. Following the focus groups, two additional participants were recruited, and their photographs and individual interview data were used to verify preliminary themes (Table 1). All interviews were recorded and transcribed verbatim.

In keeping with the tenets of PAR and photovoice (Wang & Burris, 1994, 1997), participants collaborated with the iterative data collection and analysis process. The first step in data collection and analysis began when participants chose what to photograph. Photographs were then selected, contextualized and coded. Participants selected photographs to discuss, and gave them meaning by describing what was captured and how and why they chose the subject matter (contextualization). KAL and SLV then used content analysis to analyze the interview transcripts; in particular, a double coding qualitative method was used to enhance the reliability of the data coding system (Miles, Huberman, & Saldana, 2014). The researchers independently coded segments of the individual interview and focus group data using words or phrases that described participants' actions or experiences. The researchers held regular meetings to discuss the coding, and a preliminary list of themes was developed by consensus. Data collection and analysis were iterative, and preliminary themes were discussed during subsequent interviews to ensure that the findings resonated with participants' experiences (Bradbury-Jones, Irvine, & Sambrook, 2010). No new themes emerged during the final two interviews, and recruitment ceased when we determined that the collected data was

sufficient for exploring our research questions. While additional participants might have generated new insights, we determined that our themes and categories provided a robust exploration of participants' experiences of living with DM1. Transcripts were re-examined then re-coded using the finalized list of themes. Themes were then consolidated into categories. The authors kept a reflexive journal throughout the study to record general impressions of emerging findings and to track theme and category development. Nvivo®, a qualitative software program, was used to organize and manage the data.

This study complies with the Helsinki Declaration of 1975 and was approved by the Western University Research Ethics Board.

4.4 Results

All participants were actively engaged research collaborators who took 0-40 photographs that captured symptom impact on their activities and quality of life, their management strategies, and their sense of self. We identified seven themes that were consolidated into two categories titled *A Shifting Identity* and *Managing Limitations*. Participants described that their shifting identities caused them to lose their sense of self and to grieve for their lost abilities; however, they were able to find solutions to cope with their evolving needs. Participants also discussed their variable degrees of study participation; while some participants had difficulty following study procedures, others were able to problem-solve strategies to mitigate challenges. Regardless, all participants provided rich descriptions about their experiences living and coping with DM1.

4.4.1 Participating in Research

In general, participants were actively engaged in the project and stated that they enjoyed participating because it gave them the opportunity to share their experiences and socialize. Some participants were either the first in their family to be diagnosed, or had never met anyone with DM1 outside of their immediate families. Consequently, the focus group was an opportunity for them to meet other DM1-affected individuals and discuss common experiences:

I meet a lot of people with MD (*muscular dystrophy*), but it's a big umbrella. So there's all different kinds. But I only know one person that has myotonic dystrophy and I'd like to meet other people that have myotonic dystrophy. Like males, females, different ages, and stuff like that. Maybe there'll be a friendship, you know? 'Cause we can talk and we understand what each other is going through, or something like that (Tim).

There were, however, challenges associated with using an innovative qualitative research approach with this population. Transportation was problematic; two wheelchair bound participants (Jenny and Frank) lived approximately two hours from the study site. While Jenny's caregiver was able to drive her, we had to arrange special transportation for Frank. As a result, Frank's individual and focus group session were conducted the same day, and he stated that the long study visit made him feel fatigued. Also, Frank's speech impairment rendered it difficult to understand and transcribe his comments. Moreover, two participants (Laura and Marjorie) did not complete the study; Marjorie withdrew citing DM1-related fatigue, and Laura did not attend her scheduled focus group meeting. Finally, two participants (Max and Peter) had difficulty using the digital camera:

I was trying to ... on Wednesday I was trying to take pictures of my bath seat and my walker. I put the camera on, I pushed it and it didn't go off, the flash stopped working. So... Well, I wasn't sure what to do, so I thought I'd just come down and tell you. Maybe I pushed the button that I shouldn't have, you know? (Peter).

However, four individuals (Tim, Laura, Marjorie, and Jenny) facilitated their participation by asking a friend or care partner to help them take pictures. Together, Marjorie and her husband created a list of her challenges and most problematic symptoms and then brainstormed scenarios to photograph these limitations. Other care partners assisted with picture taking and/or participated in the individual interviews. All participants described their interest in participating in research in general—and this project in particular—because it gave them the opportunity to educate researchers and clinicians about their lives.

It gives them an idea of what it's like and learn that it's not all negative and that ... like I say, you don't know what it's like unless you live it, but to take pictures

you can see what my life is like. Not all negative, but not all positive either.
Yeah, it's a good idea (Frank).

4.4.2 A Shifting Identity

Participants described that their DM1 diagnosis caused an identity shift; that is, participants' symptoms altered their physical appearance and challenged their abilities to be employed, complete household chores or participate in recreational activities. Consequently, progressive disability impacted their self-esteem and caused participants to lose their sense of self and grieve their pre-symptomatic identities.

4.4.2.1 A Changing Body

Participants' evolving physical appearance triggered their perception of a shifting identity; in particular, participants stated that their altered appearance and progressive functional decline challenged their self-confidence and their social participation. Participants commented that their changing bodies,—including low muscle tone, drooping eyelids, premature balding and facial atrophy—coupled with their need for assistive devices, significantly impacted their self-esteem. Consequently, participants worried that they were unattractive, and that their functional decline made them feel older than their chronological age:

I can't wear high heels 'cause I fall off them! I gotta buy shoes with a low heel and that makes me feel so *stupid* because I feel like an old lady. You know, with a walker I feel like an old lady. I'm not—I'm 51—but that's not old to me you know? (Laura)

Even participants in stable and loving relationships experienced poor self-esteem; some participants were not only embarrassed by their physical appearance, but also by impaired mobility and their propensity for falls: “It's not so much the scrapes on your hands and knees, it's the scrapes on your *dignity*...” (Meg). Further, participants felt judged and believed that members of the general public lacked empathy and treated disabled people unfairly. When asked to comment about her shifting identity, Jenny

stated: “Thank God I knew people when I was still *okay*—walking around and stuff—because otherwise I think I'd probably be treated differently, being in a wheelchair, people not knowing. Personality doesn't change because you're in a wheelchair.”

Similarly, other participants believed that members of the general public perceived them as slovenly or cognitively impaired, particularly if they had a speech impairment or fell in public: “I fall, they judge me as clumsy, and when I appear with mud on me because I've fallen, they assume I don't bother bathing regularly. So ...that bothers me, the people that assume” (Meg).

4.4.2.2 The Challenges of Everyday Activities



Figure 4-1: : Improvising to Manage the Challenges of Everyday Activities: Using a Rubber Grip to Open a Door

I find sometimes, especially if it's cold, if I'm cold, they (hands) don't always work, like

when they seize up. And some door handles, the outside doors are easy, it's just a push latch, it's fine, which I like better... But sometimes it's just too damn hard. (Joe)

Symptoms including weakness, gastrointestinal problems and chronic respiratory failure impeded participants' from fully participating in activities they once enjoyed. In particular, participants with diarrhea were concerned about incontinence, and individuals with breathing difficulties found it cumbersome to travel with their bipap machines. Participants described that DM1 symptoms also impacted their ability to do activities usually taken for granted like bathing, walking a short distance or climbing stairs: "I don't like stairs... Walking down stairs is fine, but going up stairs... It's hard on my legs, eh? I go up two flights and I breathe hard... Yeah, and I have to sit there and wait for a while. I sit there for a while, and then we'll go up two stories. I've got five flights of stairs to go up" (Peter). Additionally, participants described having difficulty exercising or opening doors (Figure 4-2) and bottles or jars. Moreover, the local environment was described as inaccessible to those with disabilities; narrow aisles, sidewalks in poor condition, and a lack of handicapped accessible entrances made it difficult to navigate these spaces:

I was going to take a picture of the mall because it's the only one that on the centre doors it has no buttons. It doesn't make sense because the bus, the Paratransit van, goes to a different door. That doesn't make sense to me because it should be on these two doors (Frank).

While myotonia, swallowing dysfunction (Figure 4-1), cardiac abnormalities and cataracts minimally impacted participants lives, fatigue was described as "a vicious enemy" (Marjorie) that exacerbated other symptoms. For example, Frank stated that his speech impairment worsened when he was tired, and excessive daytime somnolence caused participants to fall asleep at inappropriate times, making it difficult to work, complete chores or socialize:

Falling asleep in the middle of conversations, it's *embarrassing* because I'll fall asleep in the middle of sentences, and it's not for lack of attention or lack of interest, it's just the myotonic dystrophy makes me tired (Meg).



Figure 4-2: Making Swallowing Safer

Well, that's (swallowing) not good now... But my jaw is not working well. I can't chew. ...I can't eat beef, steak or, yeah, the chewing is a problem and I just get tired of chewing at the table and I stop...You know, I manage to have ground meats, but even so, I don't chew them. I just mush them around and swallow and I know that's not good but that's all I can do. (Marjorie)

4.4.2.3 Loss and Grief

Participants' functional decline and physical appearance left them "constantly grieving" (Marjorie) their pre-symptomatic identity. Others described grieving for lost dreams, opportunities, and their previous physical abilities. DM1 was isolating; some participants had few social connections because their symptoms precluded full-time employment or participation in recreational or leisure activities. Consequently, participants' relationships were impacted by limited finances, fatigue or a lack of motivation to be

social. Tim and Frank perceived that their disabilities were off-putting to potential romantic partners, and some participants mourned their childless state due to infertility or because they feared transmitting the gene. Ultimately, participants grieved who they had been prior to their diagnosis, and who they might have become had they not inherited DM1:

All through high school, I wanted to be a dancer. I used to take dance class and vocal class because I thought I was going to make it big and be discovered one day. There were times when I couldn't do stuff and they didn't understand why I couldn't do it. My flexibility was going and that was probably early signs of MD. Now, there's no way I could do that now, no way at all. I couldn't stand on a stage for long periods of time, dance or anything like that. There's no way (Tim).

4.4.3 Managing Limitations

Despite mourning their lost opportunities and progressive functional decline, participants were resilient and proactive about finding solutions to manage their limitations. Participants tempered their concerns about the future by adjusting their self-perceptions; that is, participants strategized solutions for mitigating their physical challenges and for finding purpose within their current abilities.

4.4.3.1 Desiring a Sense of Purpose

Participants expressed the need to feel valued and to be seen as contributing members of society: "Sometimes I'll sort it (*laundry*) upstairs and that way at least they know I'm doing *something* ... (Laura)." Many described desiring a sense of purpose and sought to maintain their independence and sense of control. Individuals took pride in their abilities and sought validation in volunteering or engaging in part-time work, being proactive about their health, driving, and completing household chores. There was a clear sense that they tried to reframe their lost identity by focusing on their achievements and current abilities. Like Joe and Frank, Max described that he had once been an award-winning athlete; however, when asked about his current accomplishments, he quietly stated that he took pride in:

Staying alive, basically. Being able to live on my own and function as a human being to do what I do. I'm able to watch TV, listen to the radio, talk on the phone and try to keep my place presentable to anybody who's going to come over.

4.4.3.2 Finding solutions

Participants described changing their expectations, creating new goals, and finding solutions for everyday challenges. In particular, individuals discovered tools or strategies to help them maintain their independence and functional ability while bolstering their self-esteem. These strategies included engaging in volunteer or paid employment, using mobility aids (Figure 4-3), taking medication for excessive fatigue, and facilitating



Figure 4-3: Mobility Aids Mitigate Functional Decline

That's my cane, my best friend. You know, it's like my third leg. I go everywhere with that cane. And I think it's great. (Tim)

household chores by using devices like rubber grips for opening jars or doors (Figure 4-2). Participants also adjusted their expectations about social and recreational outlets:

It's there, it's going to just get worse, but it hasn't really changed my lifestyle. Other than, yeah, less exercise, less sports ... But I mean participating, even kicking a ball around with the kids. *I can't do it.* Which is pissing me off, but you get over it, you know, and do something else. I can still play cards. (Joe)

Participants also coped by moving to accessible or community oriented housing, appreciating humor, and having pets. To preserve a “non-disabled” sense of self, some participants were selective about disclosing their diagnosis. For example, Tim's friends and family were aware of his condition, but he stated that he did not take his cane to work because he worried that his co-workers would treat him differently.

4.4.3.3 Family Dynamics

However, participants relied on family members or friends to assist them with completing tasks and maneuvering the local environment. Strong family, friend (Figure 4-4), and marital relationships seemed key to living successfully with DM1. Individuals with a strong support system described having more coping strategies (particularly if there was financial stability or someone to help with chores) and feeling less isolated (particularly if they lived with their spouse or children). Overall, there was a sense that supportive family and community members eased the burden of disease and made individuals feel safe. When asked what information would be important for researchers and health care providers to know, Marjorie replied:

To fight depression. Because she's (*neurologist*) asked me about that. My doctor too. But I am not depressed because I have that man (*husband*) in my life. I couldn't be depressed.

Conversely, patients who lived alone, were unemployed or lacked family support described feeling bored and isolated. Limited finances made it difficult to participate in recreational activities; therefore, watching television was the primary leisure activity for



Figure 4-4: Supportive Relationships are Key to Living Well with DM1

That's in front of our building. We have a little pavilion. We sit on the benches and chat, and socialize, there. That's just off to the side, it's just another table where we socialize a lot. It was cold that day, so no one was there. But usually there's seven or eight people outside, chatting. (Frank)

several participants. Moreover, family or friends could exacerbate participants' feelings of isolation, disability and dependence. Tim's gene negative sister would not acknowledge his DM1, and some non-affected family and friends did not understand the limitations imposed by fatigue and weakness. For example, Peter felt pressured to participate at a level that was discordant with his symptoms:

That's part of the myotonic dystrophy, it makes you tired, eh? Mom tells me I should be out longer. I say, Mom, you haven't got it. I'm *tired*. She says, 'well,

you shouldn't be tired.' 'Well, I am, Mom.' You know, I'm almost 50 years old, I get tired.

4.4.3.4 What will the future hold?

Participants were resilient despite worries about what the future might hold as their condition progressed. Many of these fears stemmed from having watched a family member decline; some participants had a 'crystal ball' mentality and envisioned that total disability was imminent and inevitable: "Probably as my disease starts deteriorating. My hands – you know, pretty soon I won't be able to do anything and I'll go to a nursing home (Peter)." Meg worried about what would happen to her son with congenital DM1 should she become incapacitated, and others were concerned about burdening family members with their care. In general, participants seemed more concerned about the effects of their decline on others, than for themselves.

Participants described problem-solving strategies to assuage their fears. In particular, participants described using bath seats and grab bars in the shower, and putting spikes on their shoes during the winter to prevent falls. Moreover, some made plans for the future—albeit reluctantly—including financial plans and medical directives. For example, Marjorie stated that she did not want a feeding tube should her dysphagia worsen. However, there was a variable amount of financial and supportive resources available to participants, and those with supportive care partners or paid employment seemed more secure about their future.

Others, however, coped by not dwelling on DM1, and described that there were others living with far worse conditions or limitations: "So you do what you can, you do what you have to deal with, there's a lot of people who have a lot of bigger deals than this" (Meg). The primary coping strategy for most participants was to "just deal with it" and to take each day as it comes:

When it changes, you've got to make the change. That's all there is to it. You can't get it back. When it's gone, it's gone. That's the way this disease works. You have to learn to function with it. You've got to do what you can do with what

you've got left. You do need assistance. You do need people around you who can help you when you need help. Don't be afraid to ask for help, that's what they're there for, utilize that. It may not be what you're used to but you have to make the change if you're going to get along with the disease. (Max)

4.5 Discussion

Shared decision making is the 'pinnacle' of patient-centered care (Barry & Edgman-Levitan, 2012) and patients are increasingly collaborating with researchers to set clinical goals and research agendas (Schipper, Dauwerse, Hendrikx, Leedekerken, & Abma, 2014; Teunissen, Visse, de Boer & Abma, 2013). Recently, a mixed methods study assessing the research goals of patients with neuromuscular disease found that patients prioritize research that explores symptomatic management and medical care, their quality of life, and educational initiatives to raise awareness about neuromuscular disease (Nierse, Abma, Horemans, van Engelen, 2013). There remains, however, a relative lack of patient-centered, qualitative research that explores the experiences of individuals living with myotonic dystrophy; consequently, there is little evidence that patients' voices are being heard, or that their clinical and psychosocial needs are being met.

To address this, we successfully used a novel and innovative qualitative research approach that explores all of the patient-identified psychosocial research goals reported by Nierse, Abma, Horemans & van Engelen (2013). While photovoice has been used with patients with a variety of chronic neurological conditions (Genoe & Dupuis, 2013; Levin et al., 2007; Lorenz & Kolb, 2009 & Wiersma, 2011) it has not been used in DM1. We found that photovoice is a useful research method for exploring the experiences of individuals living with DM1; our research participants captured a range of experiences including symptom impact on their quality of life, their concerns about the future, and their coping strategies. While the qualitative approaches used in previous muscular dystrophy research provide rich data, we believe that photovoice is particularly beneficial for providing a unique perspective of patients' lives while giving them the opportunity to

collaborate in research. Participants' photographs literally provide clinicians and researchers with a 'picture' of patients' lives that would not be elucidated during a clinical encounter.

There were, however, limitations and challenges to using this innovative method with this population. Some participants had difficulty using the digital camera, and two participants did not complete the study. We speculate that Max and Peter's hand weakness or myotonia made it difficult for them to hold the camera and depress the shutter; in turn, they were unable to problem solve a solution for taking pictures. Moreover, DM1 symptoms including fatigue, speech impairments, mobility problems and apathy may have further impacted participants' ability to comply with study procedures. However, while DM1-related symptoms complicated participation, the benefits of photovoice far outweighed the challenges. We suggest that modifications or adaptations may be necessary to facilitate participation for some individuals. For example, while we gave participants the opportunity to practice taking pictures, we suggest that more time should be spent during the camera orientation session to clarify instructions and verify that participants are comfortable using the camera. It may also be advantageous to present participants with scenarios and then discuss potential problem-solving strategies should the camera fail to work, or fatigue or mobility impairments hinder their ability to take pictures. Finally, we did not set out to include caregivers as research collaborators, but our research participants identified them as a valuable strategy that facilitated their study participation. Therefore, it may be helpful to include caregivers at the outset of photovoice projects exploring patients with complex needs. Despite challenges, participants were enthusiastic research collaborators who were candid about the impact of their progressive symptoms on their self-esteem, their social participation and their relationships.

Therefore, this study provides a rich exploration of the impact of symptoms on DM1 patients' lives, and illustrates patients' shifting identities, a concept that has not yet been fully articulated in the DM1 literature. In particular, our study participants emphasized the impact of their changing bodies and their functional decline on their self-esteem and their sense of self. Our findings suggest that symptoms like dysphagia or cardiac

abnormalities are not overly troubling for participants despite their propensity to cause sudden death (de Die-Smulders et al., 1998; Garrett, DuBose, Jackson, & Norman, 1969). Instead, participants were troubled by symptoms that directly impacted their quality of life like weakness, fatigue and their altered physical appearance. These findings resonate with other muscular dystrophy patients (Boström & Ahlström, 2004; Cup et al, 2011; Gagnon, Mathieu & Noreau, 2007; Heatwole et al, 2012; Nätterlund et al., 2001), and clinicians' and patients' divergent goals are well-described in the neurological disease literature (Bonelli & Hofmann, 2004; Heesen, Kopke, Richter, & Kaspar, 2007; Martinez-Martin, Rodriguez-Blazquez, Kurtis, Chaudhuri, & Group, 2011). In particular, a study of caregivers for individuals with DM1 and dysphagia found that weakness and fatigue are more concerning to caregivers than swallowing dysfunction despite the potential for choking to lead to aspiration pneumonia or sudden death. It is likely that the caregivers' concerns mirror those of their affected family members (Authors, in press).

Study participants also described the impact of DM1 on their relationships and discussed their feelings of isolation and boredom. Participants articulated that the genetic and progressive nature of DM1 caused them to mourn for lost abilities and opportunities. This sense of grief and loss permeated the photographs and interview transcripts, whether it was the death of a child with DM1 or the progressive loss of function and social outlets. Grief resulting from being childless—due to infertility or fear of transmitting the gene—was echoed by participants in a qualitative study examining the experiences of 46 individuals living with a hereditary muscle disease (Boström & Ahlström, 2005). In addition, some of our study participants also expressed a sense of failure or self-blame for their infertility or for giving birth to a child affected by congenital DM1. Finally, while mourning the loss of the pre-diagnosis self is a common theme for individuals with muscular dystrophy--particularly in regard to embarrassment or self-consciousness about one's appearance (Boström & Ahlström, 2005)—the extent and complex nature of DM1 patients' grief over their progressive functional decline has not yet been described. We posit that our participants experienced a cyclical pattern of loss: disability caused the loss of independence through reduced recreational opportunities and employment. In turn, these losses caused financial and social restrictions that resulted in greater social isolation

and loss of independence. Ultimately, these losses contributed to a decreased sense of self that was attributed to disability, thus perpetuating the cycle.

However, participants were resilient; their strategies for coping and maintaining independence were similar to those reported by other research participants, and included doing chores or other activities at their own pace, or re-framing their expectations for personal, social or recreational goals (Boström & Ahlström, 2004). In the present study, while participants reflected on their past accomplishments and their present limitations with some degree of sadness and frustration, they demonstrated resilience by focusing on their current achievements. Moreover, contrary to the established literature regarding the DM1 personality profile (Delaporte, 1998; Meola et al., 2003), several of our participants demonstrated initiative and showed resilience by putting their condition into perspective and taking it “day-by-day”, having a good knowledge base about their condition and an appreciation about prognosis, finding solutions to challenges, and being attentive and engaged study participants. Three participants were employed (paid or volunteer) and two were advocates for DM1; one is a leader in the DM1 community, and another chose to attend a first-year Masters of Occupational Therapy course to discuss five of his pictures and answer questions about living with DM1. The latter was in keeping with the tenets of photovoice to disseminate information to ‘stakeholders’. Our results demonstrating participants’ advocacy, perseverance and insight into their condition are significant in light of the current literature that focuses on the apathy and limited cognitive capacity affecting those with DM1.

4.6 Recommendations/Future Directions

Results from this study suggest that our study participants with mild to moderate DM1 are proactive about finding solutions for challenges. Moreover, their photographs offer a window into participants’ lives that might not be accessible during a clinic visit. Therefore, the participants’ photographs of barriers and facilitators to living successfully with DM1 might inform clinical recommendations, particularly suggestions for mobility aids or devices for assisting with daily tasks. It may also be helpful to use

the photographs to create handouts or a poster of assistive devices or mobility aids for patients to use as a reference guide.

This study identified a need for an in-depth, examination of facets of living with DM1 including isolation, grief, and opportunities for social and recreational pursuits; qualitative research is well-suited to these inquiries. Finally, since participants in the present study considered symptoms that clinicians provide rigorous surveillance for (e.g., dysphagia or cardiac abnormalities) to be minimally impactful, it would be valuable to interview health care providers and patients to ascertain which symptoms are of personal and clinical significance.

This finding also underscores the importance of, and need for, a multi-disciplinary team that is situated in either a neuromuscular, rehabilitation or a family practice clinic. In order to holistically address patients' complex needs, it is imperative that the team includes physicians, nurses, occupational and physical therapists, speech pathologists, and social workers. Nurses are educated specifically to collaborate with patients and their family members to address physical and emotional issues; in turn, nurses can help patients and caregivers navigate the healthcare system and obtain appropriate care in the hospital or in the community (personal communication, Wilma J. Koopman). Nurses, therefore, are well-suited to providing holistic, patient-centered care for DM1 patients' complex needs. In particular, nurses have the skill set to address patients' and caregivers' educational and psychosocial concerns while monitoring their symptoms and treating complications. Research suggests that patients who attend nurse-led chronic disease clinics have better self-care behaviors, improved outcomes, and greater satisfaction with their care (Hill, 1997; Strömberg et al, 2003). There may be a unique opportunity for nurse practitioners to care for patients with a chronic and progressive disease that may not be of interest to some medical practitioners, particularly when their clinic time is limited and compensation is fee-for-service (personal communication, Wilma J. Koopman). While researchers have proposed a DM1 management model and a nurse-led integrated clinical care pathway (Chouinard et al, 2009; Gagnon et al, 2007), these have not yet been systematically studied or implemented across clinical sites. More research is

therefore needed to explore HCPs and patients' perspectives of—and expectations for—a patient-centered approach to clinical care.

4.7 Limitations

This research study explored the experiences of nine individuals living with DM1 and is therefore not generalizable to a wider population of patients. However, the results may resonate with DM1 patients in other clinical settings, and findings from this article may be useful for generating new research questions.

4.8 Conclusion

Photovoice offers an in-depth exploration of patients' experiences that may impact clinical care. While it is important to consider the unique challenges presented by those living with DMI, photovoice is an informative and appropriate research method for exploring their lived experience. In particular, the participants' photographs and stories offer insight into patients' lives that would not typically be ascertained during a clinical encounter. Further, incorporating patients' knowledge about DM1 and their strategies for disease management may be useful for guiding educational initiatives and clinical recommendations. Finally, we argue that an awareness of—and appreciation for—patients' lived experience contributes to better patient-centered care, and that photovoice is useful for uncovering patients' illness experiences and generating research questions. Nurses may be particularly well-suited to addressing DM1 patients' complex biopsychosocial needs, and we propose that current physician-led DM1 clinical care models warrant further investigation.

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Chapter 5

5 Truths and Misinformation: A Qualitative Exploration of Myotonic Dystrophy Type 1

5.1 Abstract

Background: Myotonic dystrophy (DM1) is an autosomal dominant, progressive, and multi- system condition that impacts affected individuals physically, socially, and emotionally. Understanding individuals' perceptions of their disease is critical to ensuring appropriate information, education and counseling.

Methods: We conducted a content analysis of findings from a larger study that used a novel, qualitative research approach called photovoice to explore nine patients' experiences of living with DM1. Participants took pictures that illustrated barriers or facilitators to living with DM1; their photographs then formed the basis of semi-structured interviews. Transcripts were analyzed and among themes, we identified one titled DM1 Truths and Misinformation that described participants' disease knowledge. Analysis revealed four categories within this broader theme: The Physical and Emotional Cost of DM1, Managing My DM1, Genetics and Me and Patients as Advocates and Educators.

Results: Findings showed that DM1 participants had good core knowledge with respect to their disease and its implications. However, each participant held as fact, fragments of misinformation that shaped decision-making and pointed to a clear need for strategies to mitigate variable interpretation of health information.

Conclusion: We conclude that there is a need for increased education and awareness about symptoms, genetic information and treatment strategies for patients, their family members, and health care providers.

5.2 Introduction

Myotonic dystrophy type 1 (DM1)—the most common adult muscular dystrophy—is a chronic, progressive and life-limiting condition for which there are few treatments and no cure. DM1 is autosomal dominantly inherited and caused by a CTG repeat expansion in the 3' region of DMPK on chromosome 19q13 (Udd & Krahe, 2012). DM1 is characterized by anticipation; consequently, subsequent generations experience earlier disease onset and greater symptom severity (Arsenault et al., 2006). In addition to distal extremity weakness, there is a variable occurrence of cardiac arrhythmias, respiratory impairment, dysphagia, apathy, cognitive deficits, endocrine abnormalities, cataracts, pain, and sleep disturbances (Udd & Krahe, 2012). The multi-system clinical manifestations may impact the physical, emotional and social function of affected individuals; in particular, DM1 affects family planning as well as other aspects of social and psychological health (Boström, Nätterlund, & Ahlström, 2005). A DM1 personality profile is emerging from the literature suggesting that those affected may have avoidant personality traits and cognitive impairment. Therefore, it may be difficult for those living with DM1 to participate in new activities or form relationships (Delaporte, 1998; Meola et al., 2003; Sistiaga et al., 2010). Moreover, a study of 200 DM1 patients living in Quebec found that affected individuals had lower educational attainment, lower employment rates and were more reliant on social assistance than the general population (Laberge, Veillette, Mathieu, Auclair, & Perron, 2007). As a result, individuals with DM1 may experience disrupted social participation and dissatisfaction with their employment status, recreational pursuits and mobility (Gagnon, Mathieu, & Noreau, 2007). Disease severity, fatigue, cognition and mood may impact DM1-affected individuals health related quality of life (Antonini et al., 2006; Laberge et al., 2013; Peric' et al., 2010).

The literature suggests that DM1 patients' variable symptom presentations may challenge care provision (Gagnon, Noreau, et al., 2007; Heatwole et al., 2012). Furthermore, we speculate that health literacy—or “the degree to which individuals have the ability to obtain, process, and understand basic health information and services needed to make appropriate decisions” (Medicine, 2004)—may be impacted for affected individuals'

presenting with cognitive impairment, apathy and low educational attainment (Censori, Danni, Del Pesce, & Provinciali, 1990). Previous research suggests that affected individuals have misunderstanding or misperceptions about DM1 (Faulkner & Kingston, 1998; Laberge et al., 2010); in particular, an interview study with 25 DM1-affected women found that while most participants had a good understanding of their DM1 symptoms, half of the sample misunderstood genetic information regarding anticipation and the potential risks associated with maternal transmission (Faulkner & Kingston, 1998). A questionnaire study of 200 DM1-affected individuals' assessing their knowledge and attitudes about DM1 found that participants were less likely than non-affected participants to understand information about—and the consequences of—inheritance patterns and clinical manifestations (Laberge et al., 2010).

Individuals with low health literacy may be unable to understand information provided by their health care providers, including treatment advice and information about appointments (Schloman, 2004). Therefore, the doctor-patient relationship may be impacted because individuals may not understand information and may therefore be unwilling or unable to ask for clarification (Peterson et al., 2011). In turn, a survey study of approximately 1,500 patients with heart failure suggests that health literacy may impact an individual's ability and willingness to comply with treatment recommendations (Peterson et al., 2011); low health literacy may be associated with poorer health status, outcomes, increased hospitalization rates (Williams, Davis, Parker, & Weiss, 2002), and an increased risk for mortality (Peterson et al., 2011). Given that DM1-affected individuals may miss clinic appointments or seem uninterested in their health (Meola & Sansone, 2007), it is essential that researchers and clinicians assess patients' understanding of health information. To our knowledge, health literacy has not been evaluated in DM1.

We speculate that patients' misperceptions and misinformation about their health may impede their ability to participate in shared decision-making (SDM) (Charles, Gafni, & Whelan, 1997). This is problematic because SDM is widely considered to be the 'pinnacle' of a patient-centered care approach (Barry & Edgman-Levitan, 2012) that strives to use patients' needs, values and goals to guide health management (Medicine,

2014). We contend that an understanding of DM1-affected individuals' knowledge and beliefs about their condition may directly impact patient-centered care approaches; that is, this information may be crucial for informing clinical care guidelines and for developing strategies for patient self-management. Therefore, exploring the health care perceptions of individuals living with DM1 is important for uncovering their understanding of DM1 to ensure that patients have the tools to proactively seek health information and manage their care.

Few studies have used qualitative research methods to explore symptom impact on affected individuals and their family members (Bostrom & Ahlstrom, 2005; Bostrom, Ahlstrom, & Sunvisson, 2006; Boström & Ahlström, 2004; Cup et al., 2011; Geirdal, Lund-Petersen, & Heiberg, 2014; Heatwole et al., 2012). We expect that an understanding of patients' experiences with—and knowledge about—DM1 will lead to better patient-centered care. We explored the experiences of individuals living with DM1 using photovoice—an innovative research methodology that uses participants' photographs to augment qualitative interviews (LaDonna & Venance, 2014; C. Wang & Burris, 1994, 1997). One theme identified from this study related to participants' knowledge about DM1. There was a mixture of fact and misperception that had the potential to impact their health and ability to make informed decisions that has the potential to influence shared-decision making. Furthermore, this information may begin to inform health care providers about the level of health literacy in DM1. The purpose of this analysis was to conduct an in-depth exploration of participants' understanding about DM1, and to identify knowledge gaps that may challenge patient-centered care.

5.3 Methods

Twenty-two individuals with mild to moderate adult onset DM1 attending an academic neuromuscular clinic were invited to participate in a study using photovoice to explore their experiences living with DM1; nine individuals ($n = 4$ females) consented (Table 1) (LaDonna & Venance, 2014). Reasons for declining participation included lack of time or interest and distance to travel. In addition, 7 individuals expressing interest in participation were lost to follow-up contact.

5.3.1 Data collection: Photovoice

Photovoice uses the visual image to document individual experience, and it is useful for conducting research with participants with low literacy or poor cognition (C. Wang & Burris, 1994, 1997). Photovoice has also been used to explore a variety of chronic neurologic conditions (Aubeeluck & Buchanan, 2006; Guerra, Rodrigues, & Demain, 2013; Levin et al., 2007; Lorenz & Kolb, 2009; Wiersma, 2011). Our photovoice study design incorporated three data collection components: a camera orientation session, an individual interview, and a focus group session (C. Wang & Burris, 1994, 1997). KAL—a graduate student who does not participate in the clinical care of DM1 individuals—conducted all information sessions and interviews with the participants.

After consenting, all nine participants attended an individual camera orientation session in which the purpose of the study was described, the ethics of picture taking was discussed (C. C. Wang & Redwood-Jones, 2001) and participants were provided with a digital camera and instructed about its use. Participants were asked to “take pictures of what it is like to live with DM1”, and to photograph barriers and facilitators to their health. Participants had 2-3 weeks to take pictures and then returned for an individual interview. Each individual interview was open-ended and participant directed; that is, all 9 participants began the interview by discussing his or her photographs. KAL would then use probes such as: “Can you tell me about DM1?” that were followed by questions pertaining to participants’ disease history, symptoms and daily activities. At the end of the individual interview, participants were asked to select 2-3 pictures to discuss during a focus group session. Five participants participated in one of two focus group sessions of 2 and 3 participants. Participant 2 withdrew from the study citing fatigue, and Participant 3 did not attend her scheduled focus group session; their data was retained and analyzed. Participants 8 and 9 only participated in individual interviews; their data was intended as a ‘member check’ (Lincoln & Guba, 1985) to ensure that the themes identified resonated with their experiences. We ceased recruitment when we determined that no new codes or themes were emerging, and that our data was therefore sufficient for providing a robust exploration of participants’ experiences living with DM1.

5.3.2 Data Analysis

Participants were active collaborators in the data collection and analysis process; in particular, participants chose—and gave meaning to—the subject matter captured in their photographs (C. Wang & Burris, 1994, 1997). In turn, an inductive content analysis (Miles, Huberman, & Saldana, 2014) was used in which the authors coded the interview transcripts using words or phrases that described participants' actions or experiences. The most frequently occurring codes were collapsed into themes and categories. Of the nine themes identified, one related to participant knowledge of their disease coded as DM1: Truths and Misinformation, captured passages describing patients' knowledge or misconceptions about the etiology, inheritance patterns or symptoms of DM1. To provide a deeper exploration of this theme, two neuromuscular clinicians (SLV and AG) reviewed and coded these passages using words or phrases that described the topic being discussed (e.g., “genetics” or “symptom impact”), then labeled them as a “truth” (i.e., ‘good’ understanding) or as “misinformation”. KAL then reviewed and consolidated the codes into categories. All researchers met regularly to discuss the codes and categories and to resolve areas of discrepancy; in particular, discrepant issues were resolved with discussion and/or by reviewing the patients' transcript in tandem with his/her chart. The final list of categories was developed by consensus. NVivo®, a qualitative research software program, was used to organize and manage the data. To enhance the study's relevance and resonance for clinicians, we conducted a retrospective chart review to collect demographic and clinical information (Table 2).

The study was approved by the Western University Research Ethics Board, London, Ontario, Canada.

5.4 Results

All patients had been symptomatic for many years (range 8 to 34 years, mean \pm SEM 19.2 ± 3 years) prior to participating (Table 2). Seven of the nine participants had a family history of DM1, and all but one were ambulatory. In general, all participants discussed DM1 symptoms and their impact on daily life with varying degrees of

certainty. Yet, all participants stated as fact, opinions that reflected misperceptions on the part of self, friends, family, family physicians, specialists and others. Four categories were identified: *The Physical and Emotional Cost of DM1*, *Managing My DM1*, *Genetics and Me* and *Patients as Advocates and Educators*.

Table 5-1: Participant snapshots

Participant 1	Mid 50s and has mild DM1. Participant 1 asked her husband to assist with data collection; together, they took 4 pictures. She completed her individual interview, but did not attend her scheduled focus group session.
Participant 2	Early 70s and has mild-moderate DM1. Participant 2's husband helped her take pictures and he attended her individual interview. They discussed 13 photographs. Although Participant 2 was an enthusiastic research participant, she withdrew from the study because of fatigue.
Participant 3	Early 40s and has moderate DM1. He took 15 pictures and participated in Focus Group 1.
Participant 4	Mid 40s and is moderately affected. He took 40 pictures and participated in Focus Group 1.
Participant 5	Early 40s and is mild-moderately affected. Participant 5 took 11 pictures and participated in Focus Group 2.

Participant 6	Mid 40s and has moderate DM1. He was unable to take pictures because he had difficulty using the camera. Instead, his interview was structured around the images he would taken. Participant 6 participated in Focus Group 2.
Participant 7	Early 40s and has mild DM1. Participant 7 chose the subject matter of her 28 images, but her partner took the pictures on her behalf. Both participated in the individual interview session, and Participant 7 was a member of Focus Group 2.
Participant 8	Late 50s and has moderate DM1. Participant 8 stated that he had difficulty using the camera; consequently he was only able to capture one image. Participant 8's individual interview was used as a member check of the preliminary findings.
Participant 9	Early 40s and is mildly affected. Participant 9 took 5 photographs, and her individual interview was used as a member check of the preliminary findings.

Table 5-2: Clinical and demographic background of the participants continued

Duration of Disease at Time of Study	19.2 \pm 3 Years
Age of Symptom Onset	27.6 \pm 4.5 Years
Time to Diagnosis	5.4 \pm 2.9 Years
Post-Secondary Education	2/9 (22%)
Employed	2/9 (22%)
Family Member with DM1	7/9 (78%)
Number of Trinucleotide Repeats	793 \pm 38
Ptosis	7/9 (78%)
Dysphagia/Dysarthria	7/9 (78%)
Respiratory Involvement	5/9 (55%)
Weakness	9/9 (100%)
Ambulatory	8/9 (89%)
Diabetes	2/9 (22%)
Cataracts	6/9 (67%)
Cardiac Involvement	4/9 (44%)
Depression	3/9 (33%)
Excessive Daytime Sleepiness	6/9 (67%)

5.4.1 Physical and Emotional Costs of DM1

All participants were aware of the variable presentation and progressive nature of DM1 and were able to accurately describe neuromuscular and systemic manifestations. Participants were often knowledgeable about symptoms and disease complications regardless of whether they had experienced them personally. In particular, patients correctly identified weakness, droopy eyelids, fatigue, myotonia, sleep abnormalities, pneumonia, diabetes, cataracts, and cardiac abnormalities as complications of the disorder.

“You got muscles. Like my heart’s a muscle and since I have heart disease, I said where’d that come from? Myotonic dystrophy they told me. And that affected my muscle and my heart so they had to put in a pacemaker to pace it. But like I said, what’s going to happen eventually? They just keep putting in a new thing every ten years or whatever and after that they just said, it just stays in until you die I guess” (Participant 1).

Occasionally, participants did not relate symptoms or consequences (e.g. excessive daytime sleepiness or ptosis) to their DM1, or they stated that potentially unrelated symptoms were caused by DM1. For example, Participant 2 identified a “cough” that was chronic—and clearly unrelated to her DM1—as the symptom that led her to believe she had inherited the disease from her father: “On my mother’s side there is no one. No one. It was dad and I know that because I nursed dad and I was the oldest girl and I was a nurse. So anyway, we spent a lot of time at their house. Dad got this cough and he would almost whoop. Well I have that...” (Participant 2).

The emotional burden of DM1 on participants was significant (Figure 1), and participants articulated their grief for lost function, limited social and employment opportunities, and the ability – largely attributed to inheritance and the understanding of genetics - to have children. Moreover, participants had insight that DM1 affected their physical appearance,

resulting in body image concerns. These concerns were magnified by perceptions that the general public's lack of awareness about DM1 contributed to judgmental behavior.

“The main problem I have with this is not something you can see because a lot of people don't understand. Like, if you don't have an arm, okay, they figure that out. You know, if you don't have a foot or you have to wear shades, fine, but you can't see the myotonic dystrophy, and people judge. But it bothers me the most with (son with DM1) because they judge a person's intelligence by their speech. Son's speech is not the clearest so they assume he's not the brightest, and he knows they assume he's not the brightest because of his speech, so that bothers me” (Participant 9).

Participants correctly perceived the likelihood of progressive disability over time, but there was a ‘crystal ball’ mentality in which participants envisioned that worst case scenarios were inevitable. Participant 8 described his concerns about needing assistance in the near future: “Probably as my disease starts deteriorating. My hands – you know, pretty soon I won't be able to do anything and I'll go to a nursing home.” Finally, some participants were aware of the early mortality that is associated with DM1 when symptoms present in adolescence or early adulthood: “Because, like people with myotonic dystrophy do have a shorter lifespan. Usually late 50's, 56 and up usually is when... Not necessarily that you hit 56 and you die. But you're still going to get worse and probably be in bed more and stuff like that” (Participant 3).



Figure 5-1: Physical and Emotional Costs of DM1: The Challenges of Everyday Activities: When asked what household chores she had difficulty with, Participant 7 responded: *Everything. Actually, vacuuming...Getting places to do the dusting and whatnot, depending on how much room there is. I can't stand for long periods. I had to get a new fridge where the freezer was on the bottom so I could reach down into it, it's a drawer type. It makes it much easier to get things.*

5.4.2 Managing My DM1

Participants understood that while DM1 is currently without treatments that slow disease or provide a cure, there are a variety of strategies available for symptomatic management. The need for multi-disciplinary care was acknowledged, and participants were able to identify dietary management, feeding tubes, BiPAP, mobility devices, and regular monitoring for cataracts and cardiac care as important strategies for managing DM1. However, some participants engaged in a variety of self-care strategies of variable efficacy:

“I am on a BiPAP machine but only during the night...I feel I need it because it is easier to breathe with it on...Because I have an air purifier, it takes all the toxins

out of the air and you get pure air and that takes away, that I don't have to wear that because the pure air is there. I still wear the thing at night but the pure air is in the rooms all where I live so it's just pure air and it's clean, it's fresh and it's good, good for the body" (Participant 6).

Furthermore, Participant 7 stated that her cough assist device improved her swallowing difficulties, and others believed that strenuous exercise was detrimental and should be avoided. Several participants, however, described that staying active could be beneficial; for instance, Participant 9 encouraged her affected children to be physically active because she erroneously believed that exercise would make them stronger, reverse their muscle weakness and atrophy, and improve function:

"You know... there's no reason why my kids can't be great at karate even though they have muscle weakness. That's part of why they're in karate is to strengthen their muscles so when their muscles do let them down, they have more muscle memory than the average so they'll buck up to normal."

Finally, some participants articulated the rationale for being unable to drive due to excessive daytime sleepiness or muscle weakness, and compensated by arranging transportation through friends and family or by using mobility devices such as scooters (Figure 2) wheelchairs, or canes.



Figure 5-2: Managing My DM1: Using A Mobility Device to Navigate Public Spaces: *That's my scooter, that's very helpful. My hot rod.... So, yeah, I just use it to go the library or just to go to downtown or whatever, the mall, around the building...it helps me a lot. (Participant 4)*

5.4.3 Genetics and Me

All participants described that DM1 was an inherited disease, but participants' knowledge about genetics was unpredictable. Therefore, while some participants were well-informed about genetics, most described some incorrect information. In particular, participants did not accurately describe genes, inheritance patterns and the relationship of gender to the disease. While these beliefs were accepted as fact, it was also clearly apparent that participants had grappled with, and tried to make sense of, this very complex topic. In an effort to describe genetics to her child, one participant likened DM1 to a 'broken stair' in the DNA structure.

“On one of the floors on (science museum) they have the double helix, so I was demonstrating to (daughter), this is what the helix looks like, and this, and I broke apart the stairs, is what ours looks like because that's where the myotonic dystrophy is, so she understands that. So that's what the myotonic dystrophy is, a broken stair in our spiral staircase.” (Participant 9)

Participants were able to describe anticipation and the severity of a childhood presentation of DM1, yet misconstructions about inheritance were common. One participant stated that the likelihood of an affected sibling having an unaffected child was “slim”, another described that there was only a 25% chance of passing the gene on, and several incorrectly described the effect of gender on inheritance: “... they say mother to son, and son to daughter. It seems to go that way. My sister thought it goes from mother to daughter. I said not. Most of the kids seem it comes from the father to the daughter and vice versa for the father.” (Participant 1)

Moreover, Participant 2 expressed surprise (perhaps understandably) that all of her children were affected because she believed that autosomal dominant inheritance meant that only half of her children would inherit the gene: “I am sorry all of my children have it. This was supposed to be 50%”. DM1 “skipping generations” was also reported; Participant 3 stated that his sister had undergone genetic testing and tested negative, yet he was concerned that his sister’s children might still inherit DM1: “Um, like they tested my sister and she’s fine... So, my sister’s fine. Don’t know if her children are going to have it but they’re too young to test right now. They gotta be at least in their teens and they’re not there yet. But they can have it”.

Participants also stated that gender influenced disease severity and progression; they believed that men were more likely to inherit and that DM1 progressed faster in males. However, one participant believed that the disease was more severe in females, yet did not attribute this to the risk of having a severely affected child with congenital onset. Knowledge of genetics and inheritance influenced decisions around family planning. In some cases, reproductive decisions were made prior to diagnosis. However, two participants decided not to have children after confirmation of their diagnosis to prevent

passing the gene to their children. Participant 6 had considered anticipation, and understood that he could have a severely affected child: “Because I was told that if had a child with my wife, it could be delivered with it (*DMI*) and so I said, ‘I can’t do that, I can’t do it, I wouldn’t do it.’ It’s unfortunate I can’t have a child but it’s just not a good idea.”

However, while others understood the concept of inheritance, they did not fully comprehend the risk:

“Like I mean apparently they’ve found the gene that does it. That’s the normal gene or whatever, ours is this. That’s as far as I know about it, that it’s mutated. If you have two of the little ones, you’re good. But I’ve got one of each, so it’s only 25% my kids will get it. It’s not like my partner has it. She has two normal ones, so it’s 75% chance he’s going to be normal” (Participant 5).

5.4.4 Patients as Advocates and Educators

Participants identified that the relative rarity of DM1 meant that neither the general public, nor most health care providers, were particularly knowledgeable about the condition. Participants took it upon themselves to take leadership roles in the DM1 community (Participant 4) or to educate themselves, their health care providers or other stakeholders about DM1. For example, Participant 3 visited a Masters of Occupational Therapy class to educate students about his experiences, and Participant 5 was aware that patient education resources were available. However, he stated that the medical language was daunting: “I mean I can read Harper’s book, but there’s stuff there that is beyond me. My doctor understood it, but then that’s his language. I don’t speak that language.” However, participants also recognized that these resources were beneficial for their non-neuromuscular health care providers, and participants depended on specialists to distill complex information:

“As I said, I gave him [family physician] that Harper book and he went to town with that, and then he found as much as he could on his Blackberry. But he had

never come across it, at least not myotonic. I'm sure he's seen or heard about MD, but not the myotonic. But, yeah, he found as much as he could" (Participant 5).

Participants also discussed that there was limited clinical or pharmacological research available for DM1, particularly in comparison to other neurological illnesses. Participants were however engaged in, and advocates for, more basic science or patient-centered research:

"Well I like to see things get better in the healthcare field for people with what I have and other people have what they have. Because like you said, there is no medication for us with this disease, they're working on that, I hope they find something. ... So I'm here to try to help them the best I can, that's why I'm here (participating in research)...it's my health. I want to do what I can" (Participant 6).

5.5 Discussion

We examined the DM1-specific knowledge of individuals who were part of a larger qualitative research study that explored the experience of living with DM1. Findings revealed that DM1 participants had an overall solid foundation of core knowledge with respect to their disease and its implications. However, each participant held as fact, fragments of misinformation that often shaped decision-making. We suggest that care cannot be 'patient-centered' if patients do not have the correct information to make informed health care decisions. Therefore, we identified a clear need for strategies to mitigate variable interpretation of health information. Our findings have implications for the health literacy of—and patient-centered care provision for— DM1; in particular, we identified a clear need to address DM1-affected individuals'; knowledge gaps to optimize their ability to participate in shared decision-making.

Study participants were able to knowledgeably describe many of the multisystem complications of DM1. It is possible that patients learn the clinical manifestations of the disease through their lived experience. Twenty-five participants with affected children had more knowledge of the disease than subjects without affected children suggesting that experience rather than counseling may drive patient knowledge (Faulkner &

Kingston, 1998). However, participants sometimes misattributed the causality of symptoms such as excessive daytime sleepiness, and used their own strategies such as increasing caffeine intake to try and ameliorate fatigue. These findings resonate with studies that have examined the knowledge and perceptions of individuals with DM1 (Faulkner & Kingston, 1998; Laberge et al., 2010). Of the 200 individuals who completed a DM1-knowledge assessment questionnaire, 30.5% believed that they had a poor grasp of disease-specific information (Laberge et al., 2010). In particular, affected individuals were less likely than non-carriers to recognize that physical limitations, apathy, learning difficulties and decreased social and employment opportunities were potential implications of DM1 (Laberge et al., 2010). Importantly, patients with a number of neuromuscular diseases considered a lack of information about their condition more concerning than their degree of disability (Abresch, Seyden, & Wineinger, 1998).

Genetics was the theme most frequently identified with inaccuracies in interpretation for participants in the present study; although we recognize that this study was distant from any genetic counselling participants might have received. Nevertheless, this finding is consistent with the literature. Faulkner et al. interviewed 25 reproductive-age women with DM1 and found that participants had sound knowledge of DM1 symptoms, yet only 56% of the subjects were able to describe the risk of transmission correctly (Faulkner & Kingston, 1998). Similarly, Laberge et al. (Laberge et al., 2010) found that half of the 200 DM1 patients surveyed did not have a thorough understanding of the mode of inheritance. In the present study, in addition to difficulties describing the inheritance pattern, there were expressed beliefs that gender influenced both inheritance and disease progression. While participants accurately described anticipation, the increased risk of a severely affected infant was not ascribed to maternal inheritance. This finding has implications for clinical care as all participants had received information from patient education resources, either in the neuromuscular clinic or from genetic counsellors after a genetics referral. However, this is perhaps not surprising given that findings from other qualitative research studies suggest that variable knowledge about genetics is common in other chronic disease populations (McKibbin et al., 2014; Saukko, Ellard, Richards, Shepherd, & Campbell, 2007).

We speculate that the intra- and inter-individual variability of disease expression within families over time with mildly symptomatic or asymptomatic relatives in previous generations contributes to the confusion regarding inheritance. Another potential explanation is an interaction between mild cognitive impairments in DM1 affected individuals and the complexity of genetic concepts, which are often not revisited in a systematic way in the clinic after initial discussions around diagnosis and inheritance. Cognitive deficits associated with DM1 have been described (Meola & Sansone, 2007; Sistiaga et al., 2010) and we speculate this may explain some of the knowledge gaps in a general population of DM1 patients. In patients with DM1, each additional 100 CTG repeats reduced the odds of answering questions on the mode of inheritance correctly by 18% (Laberge et al., 2010). Subtle cognitive impairment, however, may remain undetected on routine clinical assessment in the absence of the application of specific assessment tools such as the Montreal Cognitive Assessment.

Regardless, the participants in the present study were active and engaged research participants who perceived themselves as educators and advocates. Participants were able to articulate a number of common disease features with good insight into how symptoms impacted their daily lives. However, it may be inappropriate to attribute misperceptions solely to cognition; instead, other factors including (1) the variable presentation of symptoms in other family members (2) a large amount of complex information being given to patients during time-limited clinic appointments, and (3) a lack of ‘checking’ on the part of health care providers to ensure that participants are retaining appropriate information may contribute to patients’ variable understanding of complex health information.

To address these factors, we suggest that patients would benefit from follow-up appointments at regular intervals with review and assessment of relevant knowledge and given the opportunity to ask and answer questions that facilitate engagement with shared decision-making. We recognize, however, that traditional, physician-led clinical care models may be impeded by system capabilities—including time-limited appointments—from fulfilling this need. Others have suggested that a more holistic or multi-disciplinary clinical approach may be useful for addressing DM1-affected individuals’ complex needs

(Gagnon, Chouinard, Laberge, et al., 2010; Gagnon, Chouinard, Lavoie, & Champagne, 2010; Gagnon, Noreau, et al., 2007). We therefore, suggest that patients would benefit from referral to nurses and other allied health professionals familiar with myotonic dystrophy, including social workers and occupational therapists, for further clarification about symptoms or general disease-specific information, to assist patients in decision-making, and to provide reassurance, particularly in regard to disease progression and genetic information. Counseling and educational interventions may improve patient knowledge in this category (Furr & Kelly, 1999). It is likely that this information sharing will be an ongoing process to ensure adequate comprehension, retention of information and application to relevant decision making over the disease course. Anecdotally, we hosted a DM1 patient education day in which patients not only had the opportunity to learn about the latest DM1-related information and research from various health care providers, but they were also able to share common experiences with others with DM1. Given that one participant in the present study found the language of patient education resources to be difficult to understand, patient education days provides good opportunity for health care providers to simplify and clarify a wide range of health information.

5.2 Limitations and Future Directions

This is a small study that reported on the experiences of nine patients living with DM1; Findings, therefore, are not generalizable, however may resonate with DM1-affected individuals and clinicians in other settings. Similarly, we did not set out to evaluate the health literacy of DM1-affected individuals; instead, findings pertaining to disease knowledge were identified during data analysis of participants' photographs and narratives regarding their experiences living with DM1. Therefore, future research questions could be directed at the health literacy assessment of individuals with DM1 that can then be used to develop interventions and patient education materials. In turn, qualitative interviews with patients would be useful for assessing the content, readability and applicability of any education or research materials. It may also be advantageous to examine current DM1 clinical care models to determine if they are meeting patients' complex physical, psychosocial, and educational needs.

Importantly, we identified that some participants felt responsible for educating non-neurology specialists about DM1. It may, therefore, be important not only to assess and clarify patients' knowledge, but also to provide in-services, one page "DM1 fact sheets" for family and generalist physicians, or to give presentations to other specialties to ensure that accurate information is disseminated to health professionals caring for individuals with DM1. Patients may be a powerful and under-utilized resource for raising awareness in the clinical setting.

5.3 Conclusion

While patients are knowledgeable about DM1, they have misinformation and misperceptions that may affect their ability to make important decisions about their health. Findings suggest that information about genetics is particularly problematic for patients, and that misinformation may influence decision-making. Therefore, future research should address the educational needs of patients; in turn, educational interventions are required to bridge these health literacy gaps and optimize the health and decision-making capabilities of patients with DM1.

5.4 References

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Chapter 6

6 “[The neurologist] is throwing you a raft”: Exploring motivations for on-going clinic attendance for individuals living with chronic, progressive and life-limiting neurological conditions

6.1 Abstract

Background: Patient-centered care may be challenged for patients with complex chronic conditions, particularly those presenting with cognitive or behavioral impairments. It is therefore essential to explore patients’ and caregivers’ perceptions about their health expectations and their clinical care. Myotonic dystrophy (DM1) and Huntington’s disease (HD)—chronic, life-limiting neurological conditions with few treatments—are suitable for this inquiry.

Methods: Constructivist grounded theory—a qualitative research methodology that explores basic social processes—informed data collection and analysis. Fourteen patients, 10 caregivers and 11 health care providers (HCPs) were purposively sampled from one academic medical centre in Ontario, Canada to participate in semi-structured interviews. Three levels of coding were done to generate themes; in turn, themes were condensed into categories.

Results: Patient and caregiver participants described continuing clinic attendance to seek expert information and social support. Medical management, altruism and reassurance provided the motivation. However, patients’ and caregivers’ motivations change over time, with clinic becoming more important for caregivers as their loved one’s condition deteriorates. Regardless, participants describe clinic as a ‘safe place’ that decreases their isolation and empowers them to actively participate in health care and research.

Conclusion: In the absence of disease-halting or curative treatments, patient and caregiver participants perceived that they derived a therapeutic benefit from the attentive care provided by expert HCPs. However, traditional physician-led medical models may be less focused on patients' and caregivers' supportive goals, and we suggest that nurse-led—or other clinical care models—warrant further investigation.

6.2 Introduction

The current health care system more readily accommodates patients with acute, treatable problems, not those with complex, chronic, and progressive conditions (Nolte & McKee, 2008). Wagner and colleagues (2000) proposed a Chronic Care Disease Model that makes chronic disease management proactive, not reactive; ideally, key elements including team-based care, patient self-management strategies, information technologies, and integrated decision making should coalesce to enhance the patient-provider relationship and improve health outcomes (Coleman, Austin, Brach & Wagner, 2009). Wagner et al. (2005) argue that a chronic disease management approach must be patient-centered; that is, care must incorporate the experiences, values, needs, and goals of the patient and his or her loved ones. Moreover, patients should be encouraged and empowered to manage their own care; to do this, health care professionals (HCPs) need to provide patients with adequate information and evidenced-based treatment approaches (Wagner, 2005). However, we speculate that patients with progressive, chronic diseases presenting with cognitive and behavioral impairments challenge models of patient-centered, chronic care. Care is further complicated by conditions that are hereditary and have few treatments available. Myotonic dystrophy (DM1) and Huntington's disease (HD)--chronic, progressive, hereditary and life-limiting neurological conditions for which there are no treatments to slow or reverse neurodegeneration--are well-suited to studying patient-centered health care delivery and expectations for individuals with complex, chronic disease.

DM1 and HD are genetic disorders characterized by progressive physical, cognitive and behavioral symptoms that may impact individuals' personal relationships and their ability to fully participate in work or leisure activities (Gagnon, Mathieu & Noreau, 2007;

Nance, 2006). As a result, individuals may have a lower socioeconomic status and a reduced quality of life. DM1, the most common adult-form of muscular dystrophy, is a multi-system condition causing muscle weakness, excessive fatigue, swallowing dysfunction, heart and breathing problems and endocrine abnormalities including infertility and diabetes (Harper, 2001). There is also a well-recognized DM1 personality profile described in the literature suggesting that patients may have cognitive and behavioral impairments including low IQ and apathy (Sistiaga et al, 2010; Delaporte, 1998; Meola et al, 2003). HD is a progressive neurodegenerative disease with similar features including involuntary movements, cognitive impairment and psychiatric manifestations. In particular, apathy and other psychiatric features including depression, anxiety and psychosis can significantly impact individuals living with HD. However, both conditions are highly variable, and symptoms can differ substantially, even for individuals within the same family. Regardless, affected individuals have a reduced life expectancy; sudden cardiac events and pneumonia are the leading causes of death for individuals with DM1 (de Die-Smulders et al., 1998; Mathieu et al., 1999), while pneumonia and suicide are the most common causes of death for HD-affected individuals (Roos, 2010). While there is no cure for DM1 or HD, there are limited therapies available to mitigate symptoms.

DM1 and HD are relatively uncommon conditions affecting 1:8000 and 1:10,000 worldwide. Therefore, few family physicians follow DM1 or HD-affected individuals, and patients typically attend specialty neurology clinics that provide diagnostic evaluations, follow-up, and referrals to tertiary health care providers (Chouinard et al., 2009). Treatment plans may include medications to treat symptoms, surveillance for progressive symptoms, and referrals to specialists to monitor complications and quality of life, or to physical therapists for mobility occupational therapy to address activities of daily living or social work to consider disability applications (Gagnon et al, 2007; Gagnon et al, 2010; Nance, 2012; Roos, 2010; Sturrock & Leavitt, 2010; Turner & Hilton-Jones, 2008). However, patients' cognitive and behavioral impairments, coupled with a lack of treatment options, may complicate clinical care; in particular, the literature suggests that individuals with DM1 may miss clinic appointments and be disinterested in their health (Meola & Sansone, 2007) or demonstrate poor adherence to clinical

recommendations (Chouinard et al, 2009). Moreover, HD patients may be unaware of their involuntary movements or the extent of their cognitive and behavioral impairments (Kremer, 2002). Given few treatment strategies, and the lack of awareness that patients may have about their condition, it is unknown why many DM1 or HD patients maintain regular follow-up at a specialty neurology clinic.

To our knowledge, motivations for clinic attendance for individuals with DM1 or HD have not been explored. However, reasons for clinic attendance have been studied in other chronic conditions including cancer (Brain et al., 2000; Thomas, Glynn-Jones, & Chait, 1997), HIV/AIDS (Bodenlos, 2007), and asthma (Van Baar et al, 2006). For example, 833 individuals with a family history of genetic cancers attended clinic to find out information about their individual and family members' risk of inheriting cancer, to participate in research, and to learn about genetic testing, screening and prevention (Brain et al., 2000). Patients with asthma attended to manage their symptoms, seek further medical investigations, and to hear information about new treatment options (Van Baar et al, 2006). However, patients also felt compelled to attend appointments that were scheduled in advance to avoid adversely affecting their relationship with their health care provider (Van Baar et al, 2006). It is important to note, however, that unlike DM1 and HD, these conditions have evidenced based treatments that may slow progression or improve the outcomes of the underlying disease process. We, therefore, sought to explore patients', caregivers and health care providers (HCPs) perspectives about why patients with life-limiting neurological conditions with few treatment options choose to maintain regular, on-going follow-up at an outpatient neurology clinic. In particular, what do they perceive are patients' health care expectations, and does the current clinical model meet their needs?

6.3 Methodology

6.3.1 Constructivist Grounded Theory

Constructivist grounded theory (CGT) (Charmaz, 2006; 2014) informed the iterative data collection and analysis process. Grounded theory is rooted in symbolic interactionism,

and it uses inductive methods to develop theory about basic social processes that are *grounded* in empiric data (Glaser & Strauss, 1967; Strauss & Corbin, 1990; Charmaz 2006; 2014).

6.4 Methods

Individuals with mild to moderate DM1 or HD (by physician report) who maintain regular clinic attendance were recruited from outpatient neuromuscular and movement disorder clinics at one academic center in Ontario, Canada. Follow-ups for HD and DM1 patients are typically scheduled every 6 months to two years; therefore, we defined that a patient maintained ‘regular clinic attendance’ if they had attended at least two consecutive appointments. Individuals were recruited using purposeful and convenience sampling; that is, since both DM1 and HD can cause cognitive problems, health care professionals at each clinic were asked to identify and approach patients who they perceived were able to provide informed consent and comply with study procedures. To facilitate participation for individuals with limited mobility, lack of transportation, or for those who travel long distances to clinic (up to 200km), KAL consented and interviewed participants directly following their clinic appointments. Health care professionals caring for patients with DM1 and HD were also invited to participate.

Fourteen patients (n=5 DM1), 10 caregivers (n=2 DM1) and 11 HCPs participated in semi-structured interviews. The HCPs included neurologists specializing in neuromuscular conditions or movement disorders, a psychiatrist and a respirologist; a nurse practitioner; two social workers; and a physical therapist. KAL interviewed most participants; however, since KAL had working relationships with several of the HCP participants, a research assistant (CP) conducted the HCP interviews. As the iterative data collection and analysis progressed, the interview probes evolved to explore themes. Charmaz (2006; 2014) suggests that grounded theory researchers engage in initial, focused and theoretical coding. KAL first read the first two DM1, HD and HCP transcripts as a whole, then coded each line or sentence using gerunds or in vivo codes that captured the meanings or actions described by the participants. Next, KAL consolidated the most frequently occurring codes into preliminary categories, then coded

the next six transcripts using these categories to determine their fit and relevance. KAL met frequently with SLR and SLV to discuss preliminary findings. The research team finalized a list of categories by consensus that KAL used to re-code the entire dataset. Throughout the research process, data within and between transcripts were constantly compared, and KAL wrote memos and drew diagrams to capture and explicate increasingly abstract ideas about the data. Memos were invaluable not only for making the emerging analysis more theoretical, but also for determining avenues for theoretical sampling to elaborate on the categories. For example, while the focus of this study was on individuals living with chronic neurological disease, it became apparent very early in the research process that caregivers were integral to clinic attendance for several individuals. Therefore, we amended our ethics application to include caregivers. While they were given the option of participating in separate interviews, all patient-caregiver dyads chose to be interviewed together.

Recruitment and data collection ceased when the researchers determined that theoretical sufficiency had been reached; that is, while gathering additional data might have provided new insights, we determined that the collected data was sufficient for obtaining a robust exploration of the participants' perceptions of—and motivations for—clinic attendance. NVivo©, a qualitative research software program, was used to organize and manage the data. This study was approved by the Western University Research Ethics Board; all participants were assigned a pseudonym to ensure confidentiality.

6.5 Results

The process for choosing to attend an outpatient neurology clinic follows a trajectory from seeking a diagnosis, to monitoring symptoms, to seeking guidance as the condition manifests and progresses. Participants described that living with an uncommon condition was isolating, and they framed their reasons for maintaining clinical follow-up at a specialty neurology clinic by describing negative experiences with former health care providers and members of the public. In essence, the clinic was a “safe place” where they felt understood and where they could seek information and actively participate in their care. Importantly, the DM1 and HD clinics were not just resources for patients living

with manifest disease; instead family members, especially partners and children who were at risk, also came to clinic for counseling and support. Sometimes this care was informal, but in some situations family members were able to access clinic staff (for example the social worker or the nurse practitioner) without the patient present. However, the motivation, emotional experience and the importance of attending clinic seemed to change over time for both patients and their family members along various points of the disease trajectory.

6.5.1 First Contact: Seeking a Diagnosis

Patients typically present to a specialist clinic for an initial consultation to seek a diagnosis (1) when there is a family history of DM1 or HD, but the individual does not know his/her gene status; (2) when individuals are gene positive but pre-symptomatic; (3) or when individuals are experiencing neurological symptoms and may or may not have a positive family history for DM1 or HD. Findings suggest that individuals often experience anxiety prior to visits because they are apprehensive about their “genetic fate” (Dr. Green, neurologist).

6.5.2 Recalling Negative Experiences

Apprehension is also heightened by the genetic nature and variable and unpredictable disease course of these conditions. Patient and caregiver participants framed their reasons for seeking regular, on-going follow-up at a specialty clinic by reflecting on negative experiences in their daily lives or during encounters with HCPs. Often, patients had been caregivers for affected family members; consequently, they had seen first-hand the “devastating” progression of DM1 and HD and the judgment and negative attention focused on their loved one. Participants described that their loved one had been mocked in public, misdiagnosed and/or prescribed inappropriate treatments. These experiences seemed to cause great anxiety for patients as they contemplated what their future might hold:

But she (mother) just had a lot of movements and stuff. So a lot of people made fun of her all the time. But coming to the neurologist back then, it was horrific to

just see what my mom was going through. And the two of us would just bawl all the way home... just because they couldn't do nothing. And oh my God, I was freaking out for myself too (Patricia, HD patient).

Participants understood that DM1 and HD are uncommon conditions; few participants had heard about DM1 or HD prior to a family member's diagnosis, and they perceived that the conditions were poorly understood by the general public and most HCPs: "I talk to some people that they probably heard about it but I don't think they know much about it. I know me, the first time I find out about it, I didn't know what the hell it was, I didn't have the slightest idea. Hell, my doctor didn't know what it was" (Lucy, HD Caregiver). Participants also described that they or their loved one were sometimes dismissed by HCPs who were perplexed about their symptoms: "His mother really wandered for, I'm going to say a good 10 years. Everybody just thought she was crazy... nobody would pay attention to her..." (Michelle, HD Caregiver).

Some participants described having to undergo numerous medical tests that sometimes resulted in a misdiagnosis or inappropriate pharmacological management. Negative experiences further impacted patients following their diagnosis. While not unique to DM1 or HD, some participants had a poor relationship with their treating neurologist which impacted their willingness to attend clinic; consequently, they either temporarily stopped attending or they asked to be referred to another specialist such as the participant below:

I wasn't followed for anything for the myotonic dystrophy because I never really had any problems, right? I mean other than little stuff and then when I started falling, my gait had changed and I started falling a lot. Then they sent me to (doctor) and (we) had a bit of a misunderstanding shall we say. I just don't like the (doctor's) bedside manner (Maxine, DM1).

Given the uncommon occurrence of these conditions, they were usually the only patient with DM1 or HD in their family physician's practice. Therefore, they stated that their family physician had little knowledge about--or experience managing--their complex

symptoms. When asked if her husband's family physician managed his HD symptoms, Michelle (HD caregiver) responded:

Our family doctor knows nothing, actually. It's a bit disturbing.... his doctor told us, 'I've been a doctor for 25 years and I've never had a patient with it and I'm not prescribing for you. Go to the other doctor'I don't think you should be shutting a door on someone who gets something that you didn't learn about. I mean, there's so much on the internet, that he could at least give him a little bit of sympathy...

6.5.3 Clinic is Experienced as a 'Safe' Place

Once patients adjusted to the initial shock of their diagnosis and had developed rapport with a specialist neurologist, most participants perceived that the clinic was a comfortable place where patients and families could seek the therapeutic benefit of expert information, reassurance and support. As Jeff (HD) and his father described, it was sometimes difficult for participants to articulate what made the clinic seem comforting: "It's just whenever I come here, it's hard to put into words..."(Jeff, HD patient). "It's like a shrine, it's like you're going to go to, it's like going to church on Sunday, it's like going to a shrine, like when he comes here he feels, I don't know, it gives you a different feeling of something I guess (Frank, Jeff's caregiver)."

Patients and caregivers seemed to experience the specialty neurology clinic as a 'safe' place staffed by experts who were aware of their limitations and treated patients as individuals, not as a disease. They seemed comforted knowing that someone was "...looking after me that way, rather than being out there on your own and all this stuff starts happening and you have nobody to turn to" (Tim, DM1). In contrast to their experiences in non-neurological practice settings or in the community, patients felt understood at clinic; that is, they did not have to explain their condition, their physical and behavioral impairments or their appearance. Participants like Doug (HD) felt comfortable talking about HD with the clinic social worker, but not with others because "they don't understand it." Moreover, some HCPs perceived that being understood at

clinic seemed to reduce some of the anxiety of living with—and the fatigue of having to continuously explain—an uncommon condition.

I think them just knowing that people understand and that they care... I think when you make that connection, even how insignificant it might seem to us because we don't think we're doing a lot... it's someone that's trying or does understand that they have challenges and that they're dealing with that, and someone that will just listen to their frustrations (Mae, allied health professional).

Seeing others with their condition was another aspect of feeling understood. While it was troubling for some participants to see patients with more severe disease in the waiting room, for others, it provided a degree of comfort and eased their sense of isolation: “I’m comfortable coming here because other people if they come here have the same thing...It just makes me feel better knowing other people have it...(Jeff, HD).” Jeff’s caregiver added, “Yeah, you just feel that you’re not alone” (Frank).

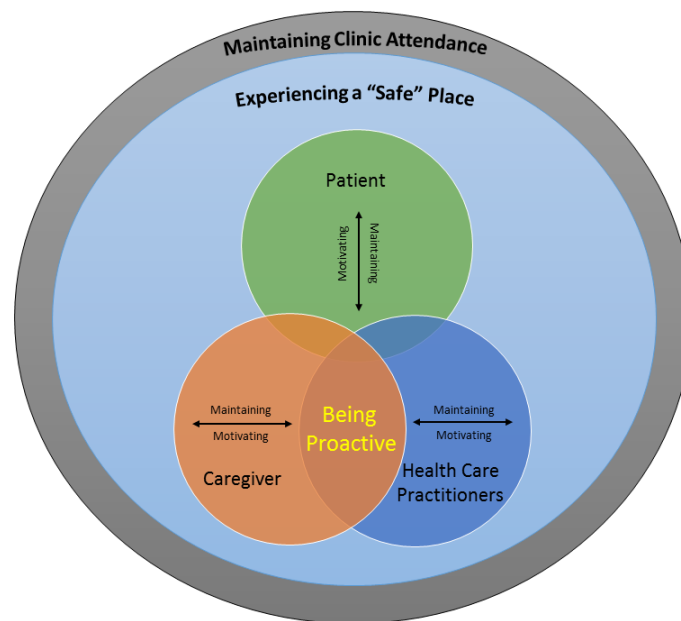


Figure 6-1: Maintaining Clinic Attendance in a ‘Safe Place’

6.5.4 Motivations for attending clinic: Seeking Expert Information

At each stage of the disease trajectory, *seeking expert information* was described as the main motivating factor for clinic attendance. “Just having a doctor or social worker or nurse who is an expert in that field, really helps them. More often than not, they have lots of questions to ask as well, which could be on a social front, medical front, nursing front, whatever. But they really have got quite a lot of questions to clarify each time that they are with us” (Dr. Night, specialist physician). Information however, meant different things at different time points, including seeking a diagnosis, education about DM1 or HD, monitoring symptoms and seeking a prognosis. Participants were particularly motivated to obtain information from experts about how their condition would impact their daily lives. Some contrasted the knowledge they received from their specialist with the care provided by their family physician:

I think since she’s a specialist per se and my family doctor is more general, like he knows maybe the idea of muscular dystrophy and what it kind of pertains to, I think Dr. Thompson would know more detail, like digs deeper into it. He might say, people with muscular dystrophy, yeah, they have trouble with their hands, but Dr. Thompson knows they have trouble with their hands but she knows why they have trouble with their hands (Tim, DM1 patient).

The clinic is also a resource for patients to seek pragmatic help completing paperwork for disability or insurance claims, and for inquiring about—or receiving assistance obtaining—medical or community resources including medical alert bracelets, support groups or funding opportunities. Clinic staff sometimes served as a liaison between patients and employers to facilitate appropriate work conditions for patients’ changing needs. Expert assistance seemed to ease these daunting tasks for patients:

Yeah, he (*Ray, allied health professional*) just happened to come out because my insurance company sent me out to apply for CPP and he offered to come out and go through it with me. So that was a big help because that was overwhelming. I just looked at it and I just put it away. ...He come right out to the house... He

wrote everything out and worded things the right way and he was awesome. I don't think I would have got it done. (Patricia, HD patient)

Patients and caregivers described that attending a specialty clinic staffed by experts was also important and necessary for obtaining the latest information about research opportunities and therapeutic advances and for having their questions answered:

I have questions that cannot be answered by anybody else, they don't know the answers. My family doctor doesn't really know the disease, I come here because I know it's helpful and sometimes you have questions that only Dr. Thompson can answer because that's what she specializes in. And sometimes, it's not only me, sometimes my caregiver will have a question for Dr. Thompson that I don't think of, for their personal knowledge or for me. It just helps me see where I'm at and if I'm having problems with something what can be done to help me, what different things, depending on what it is (Jackie, DM1 patient).

Another key reason that individuals maintained regular follow up was to have their symptoms monitored and to measure their disease progression. Often, seeking information about disease progression provided patients with reassurance that they had not declined substantially or that their current symptoms may be improved with medical management. For patients and caregivers, this was described as 'hearing good news' that relieved some of the tension of living with a progressive and unpredictable illness:

...one day Martha was testing my strength and she was doing my shoulders and she told me, she goes, for somebody with MD, you have very strong shoulders. That makes me feel good because I know, okay, I have strong shoulders at least. Something is strong on me, right? So, I think having them as kind of a safety net or knowledge of information or just something to fall back on is a good feeling I think. Everybody needs something. (Tim, DM1 patient)

6.5.5 Seeking Connection

Given the inherited and unrelenting physical and cognitive decline of DM1 and HD, participants described experiencing shame or stigma, and in turn, several felt compelled to ‘hide’ or keep their diagnosis a secret. Participants described that they did not tell employers about their condition and avoided friends and family; others had family members who refused to acknowledge or speak about the disease publicly. Participants feared that their genetic status could impact them financially (job or insurance loss) or socially (avoidance; fear of being mocked). Isolation and a sense that patients were ‘fighting the battle alone’ permeated the interviews. Therefore, the lack of community awareness was exacerbated—and patients’ sense of isolation increased—by an unwillingness for some individuals with DM1 or HD to share their stories: “I would never go over to your house and just sit down and start chatting about what I have, it just wouldn’t happen, so it (clinic) kind of makes you kind of talk about it, even though you may not want to, but it needs to be addressed” (Beth, HD patient).

Clinic, therefore, gave patients and caregivers the opportunity to form relationships and share their stories with HCPs and other families living with these conditions. For some, clinic was viewed as a social outing where they could meet—perhaps for the first time—others with their condition, or form relationships with HCPs who were understanding and who specialized in their uncommon condition:

Each day goes by and you’re wondering, where is this disease going to end up? And then, seeing the specialists that know what they’re doing helps to give you some hope. You see somebody that understands, that can understand the Huntington’s, or whatever, somebody that wants to take interest in it...(Rob, HD patient).

Participants described that their HCPs were “easy to talk to”, and some described their HCP as a friend. Participants appreciated ‘small talk’ during clinic visits about personal hobbies or interests, important life events like weddings, and the sense that they were known and treated as an individual, not as a disease: “I don’t know how they all know every little detail about me ...” (Patricia, HD patient). A sense of connection seemed

equally important for the HCPs: “I do love it and I don’t need to be doing this job any longer... but I’ve connected so well with the Society and with the families that I work with that I feel part of their family now” (Ray, HD social worker). HCPs also described that it was a “privilege” to care for multiple generations of a family (Dr. Green, neurologist), and they enjoyed forming warm, long-term relationships with patients and their family members and helping them navigate complicated decisions (Dr. Matthews, neurologist).

6.5.6 Drivers of On-Going Clinic Attendance

Seeking expert information and connection are driven by three factors: symptom management, altruism and hope. While there are no treatments to slow or reverse neurodegeneration, there are management options available to address problematic symptoms including medications, assistive devices (e.g., mobility aids; BiPap), referral to specialists, and counseling. For some patients, however, seeking a tangible pharmaceutical treatment was not their main goal for attendance. Patients and caregivers either perceived that there were few treatments available, or they were not aware that medications or interventions that their clinician had prescribed (e.g., psychiatric medications for behavioral complications) were treatments for their HD or DM1. HCPs also indicated that pharmacological treatment was not always the patients’ and caregivers’ motivation for attending clinic:

From our point of view, we want to make sure that they are getting the right treatment. But, for them, treatment is just a part of the management. The physician is very interested to improve the affective symptoms, for example, from a psychiatry point of view, then you are just very interested in controlling the motor symptoms. But the patients, all they want is to make sure they get the best treatment and that’s why they come back. And things change as well, they’re always evolving (Dr. Night, specialist physician)

Instead, altruism seemed to be the main driver for maintaining regular follow-up. Patients and caregivers described that they wanted to be advocates for individuals living with their condition. Some participants described that they believed that little could be

done to help them, but that their regular attendance at clinic might help future generations, primarily their children or other at-risk family members. In particular, participants came to clinic to hear about—and perhaps participate in—therapeutic advances and research opportunities; for instance, one participant with an unusual disease presentation consented to having her blood sent for analysis, and several other participants had enrolled in clinical trials.

Patients viewed themselves as educators; that is, they provided information to their non-specialist HCPs, and they perceived that by attending clinic and participating in research, that they were teaching consultants and trainees more about the condition. In turn, their participation would raise greater awareness for the medical community that could lead to therapeutic breakthroughs. “I like to participate with (*researchers*) so people learn about it, so maybe somewhere down the road there’s a cure or some time down the road there’s something that people will learn and then, like after I’m gone, people that have it may have it easier than I do basically” (Tim, DM1 patient).

Most HCPs indicated that their main role was to give patients and their families hope by providing management strategies and information about research and therapeutic advances. For some, attending clinic gave patients and their families something to ‘cling’ to, namely reassurance that the patient’s symptoms were being managed and that the medical and research communities were working toward a therapeutic treatment or a cure:

It’s kind of like you’re in a lake, drowning. And he’s throwing you a raft, while other doctors are just standing around doing nothing. At least Dr. Green is throwing us a raft, some kind of a dingy. Some hope that, if you work hard enough, you might be able to get on the raft and float away (Thora, HD caregiver).

However, while symptomatic management and altruism provided hope for some participants, this experience was mixed. Others sensed that they faced progressive decline with few treatment options to reverse or mitigate deteriorating symptoms, therefore they believed that “hope is what you make. I don’t depend on anybody else for

it...”(Maxine, DM1 patient). Instead, some individuals described that they attended clinic to seek social support and information, not hope:

Hope, well, I think I’m going to die with Huntington’s and not of Huntington’s...I’m not in a rush to cure me. So, if people are sort of younger, got children, then the hope is somewhere there’s a tablet that’s going to cure this. I can see why people would go for hope. I never even ask it of the doctor. I just figure Dr. Green will tell me when there’s a tablet he wants to put me on. But right now, there isn’t one available that will slow it down because it’s (*disease progression*) slow enough as it is. I don’t know. I could see the hope. I don’t go for hope, no, I go for information, definitely not hope. Maybe in 10 years I might say, oh well, maybe they’re going to do something (Margaret, HD patient).

6.5.7 Evolving Motivations

However, patients’ and caregivers’ motivations for attending and their degree of participation in their health care changed over time; immediately following diagnosis, some caregivers had to prod their loved one to attend. However, once patients had adjusted to their diagnosis, attending clinic generally became less anxiety provoking, and patients typically became active and enthusiastic care participants:

He didn’t want to go...it was a fight, his girlfriend at the time and myself had been trying to get him to go because we knew he needed to. And finally, I was so worried he would cancel the day we finally were to go ...Once he met Dr. Green and Ray, everything was great.... he just felt very comfortable with both of them (Sue, HD caregiver).

Some participants indicated that, over time, the need to measure progression became more important for caregivers than for patients because this information helped caregivers manage uncertainty and provided guidance about seeking different care or resources for their loved one. For instance, several patients were not interested in knowing how their condition was progressing, while caregivers acknowledged that

having this information was important for considering future care responsibilities as one caregiver discusses her:

And we want to be prepared ... we had a lot of stuff for my mom with, like, a walker and ... we have lots of bars for him. And Dr. Thompson mentioned the community help at ... So I think it's us that are more seeking the information than Nick because he just takes every day at a time (Vanessa, DM1 caregiver).

Moreover, as the patients' cognitive and behavioral function progressively declined, caregivers once again took an increasingly active role getting patients to clinic and directing the clinical encounter. Caregivers were a source of information for clinicians, particularly if the patient was unaware of—or downplayed—his or her symptoms:

So I think from a patient's perspective, they walk in there presenting the best that they possibly can and as much as they're coming for a reason, they don't want to talk about those reasons. And the caregiver comes with, 'and here's what's really happening' and 'this is why I'm here today'. So I think that there are two different agendas that are always happening when two people come into that room together. (Ray, HD social worker)

In the end stages of disease, while visits still addressed symptom management for the patient, the focus of the visit often shifted to supporting family members as they coped with their loved one's complex and progressively worsening condition. In a sense, as the patient's condition deteriorated, 'caring for the caregiver' often became the goal of clinic visits.

6.5.8 Taking Charge—Encouraging a Patient and Family Centered Approach

Despite their motivations for attending, participants acknowledged—and HCPs encouraged—that patients and caregivers were responsible for taking charge of health care decisions and directing care. Sometimes caregivers would speak for the patient—particularly as the patient's cognition or speech became increasingly impaired—but

HCPs would continue to encourage the patient to speak for him or herself: “It was funny because we were answering questions for Nick and she (Dr. Thompson) goes, I know you love him and you’re his care-keeper but I would like Nick to talk” (Vanessa, DM1 caregiver). Moreover, when asked about her goals for clinical follow-up, the nurse practitioner stated that the focus of the clinic visit was to address patient-identified needs: “I don’t have any goals, it’s whatever the patient determines is their goal. 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.” HCPs typically used the clinical encounter to identify worsening symptoms in order to manage and prevent complications, but a psychosocial approach sometimes usurped the traditional medical model if patients identified concerns they wanted to address:

(Describing a typical dictation): ‘You’ll see that today I didn’t examine him or her because we spent all the time...talking about their difficulty with weight loss, eating properly and making sure that blah, blah, blah.’ So, not necessarily did I have to examine their reflexes every time, because we spent 15 minutes...talking about personal issues...’ (Dr. Bennett, neurologist).

Patients and caregivers also decided if, when and how much information they wanted to obtain about their condition. For some, reading about their condition or attending a support group was beneficial. Others, however, were determined to take it ‘day by day’ and not research what their future might hold, or they did not feel emotionally prepared to read about the potentially devastating complications of HD or DM1:

Well I know what it is and I know what it does and stuff like that but for me to learn more about it I have to be in a certain state. I don’t like to read about it too much, I have to be in a certain frame of mind to read about it, because it’s not exactly the most enjoyable reading material, I have to be in a certain state to read about it and learn more about it (Jackie, DM1 patient).

For most patients, the clinic was enough of a supportive resource that they did not feel inclined to seek further support elsewhere. Some patients and caregivers were opposed to

the idea of attending a support group; participants were either uninterested in the topics presented at group, or they were not ready to see individuals with more severe disease. Reasons for choosing to obtain information or support varied for patients and caregivers:

I think it probably would be a great idea if Rose went and talked to people about that kind of stuff because it's just going to get worse. It's not getting better obviously. And me telling her or saying it upsets me, it doesn't do anything. But maybe if she heard from other people, maybe how they deal with somebody like me. That's how I dealt with my mom, but that's just me. You have to do your own thing, right? But maybe the support groups, not with me. I'm not ready to go to it. But I know one day I want to, but not right now. But maybe it would make you feel better (Patricia, HD patient).

6.6 Discussion

We identified that patient and caregiver participants attend clinic to be proactive seekers of expert medical management, community support, and information about DM1 or HD; these factors are driven by their motivation to manage symptoms, decrease their isolation, and learn about their uncommon condition. Researchers who used an exploratory qualitative approach to explore the experiences of 9 individuals (n=1 DM1) attending a neuromuscular rehabilitation centre in the United Kingdom also found that information seeking and a sense of community were important factors for clinic attendance (Hartley, Goodwin & Goldbart, 2011). However, our study advances the work of Hartley, Goodwin and Goldbart (2011); that is, the authors' study explored the experiences of attending clinic for patients with various nerve and muscle conditions, but our study adds descriptions about what motivates individuals with DM1 and HD to present to clinic, and the factors that facilitate their desire to maintain follow-up. We also captured the perspectives of caregivers and health care providers regarding on-going clinical follow-up for DM1 and HD. Patient and caregiver participants' describe that attending clinic was beneficial because of the attentive care delivered by DM1 or HD experts. We suggest, however, that a different clinical model might more efficiently address participants' educational and psychosocial support needs.

Participants framed their reasons for maintaining on-going follow-up by describing prior negative experiences that they— or other affected family members—had with health care providers or members of the general public. In particular, participants perceived that the general public and most HCPs lacked knowledge about DM1 and HD; consequently, participants described feeling judged, shamed and isolated. At clinic, however, participants described that they felt understood and validated by experts who were interested in their condition. This is similar to findings from Hartley, Goodwin & Goldbart (2011) who suggest that the clinic becomes a place of “empathy” (p. 1022 & 1029). Ultimately, the actions, motivations and drivers of clinic attendance seem to indicate that—in the absence of pharmaceutical treatment to change the underlying disease process—patients, caregivers and HCPs perceived that attending clinic was, in and of itself, a form of ‘treatment’. Patients and caregivers indicated that the act of being monitored by an expert HCP meant that they were being proactive about their health, and actively participating in the “fight” to find a cure. Attending clinic gave patients and caregivers the opportunity to be advocates and educators by participating in research and educating clinicians about their condition. Additionally, patients and caregivers discussed that *seeking connections* with other patients in the waiting room or with HCPs with a specific interest in their condition was therapeutic because they felt understood and cared about. In particular, the belief they were seen as an individual, and not a disease, was key to experiencing this benefit.

Hartley, Goodwin and Goldbart (2011) posit that the specialist care provided at a multi-disciplinary neuromuscular centre in the UK might be a source of hope and optimism for patients. For some participants, the therapeutic benefit of seeking information and connection was experienced as hope, but this was not universal. Furthermore, treatment and research did not appear to be participants’ primary motivations for attending clinic. Instead, the rapport that patients had with HCPs fostered their ability to talk about their condition and to seek social support. Clinic attendance seemed to ease participants’ isolation and reassure them that the medical community had not forgotten them.

Attending clinic provided patients with the opportunity to be proactive in managing their unpredictable health. Patients with chronic and progressive conditions may feel

disempowered by their progressive functional decline, which contributes to a sense of insecurity or to a ‘disrupted’ or shifting identity (LaDonna & Venance, *in press*; Aujoulet, Luminet & Deccache, 2007). Moreover, patients living with hereditary illness may feel powerless based on the fear they may have passed the gene to their children (Aujoulet, Luminet & Deccache, 2007). Our participants’ narratives were threaded with suggestions that they had experienced some degree of disempowerment, but coming to clinic appeared to be a way for them to exert some control. Participants indicated participating in disease surveillance by attending clinic with expert HCPs meant that they were being proactive about their health. That is, clinic attendance was perceived as a tangible way to ‘do something’ to address their complex and evolving needs. We suggest that patient and caregiver participants perceived that they were collaborators with HCPs in trying to find answers for their uncommon disorders, and were therefore active, not passive recipients of care. We propose that patients and caregivers attend clinic—not necessarily for hope—but to be reassured that they are actively ensuring that the medical community has not forgotten—nor will forget-- their rare condition.

That patients with DM1 and HD are interested and engaged in their health is a novel finding considering that the literature—particularly with respect to the DM1 population—suggests that patients are disinterested in their health (Meola & Sansone, 2007) or adhere poorly to treatment recommendations (Chouinard et al, 2009). This has significant implications for patient-centered care provision in these populations; in particular, our analysis suggests that patient and caregiver participants are proactive about seeking and providing information and support throughout the disease trajectory. While HCP participants wished to be patient-centered, they indicated that this approach was complicated by patients’ progressive cognitive and behavioral decline; in particular, they described that patients become less aware of their symptoms and the importance of managing potential complications. Moreover, previous research suggests that issues of clinical concern to HCPs may not be what patients or caregivers prioritize (Authors, 2014, Heatwole, 2012; Kremer, 2002). We speculate that time-limited appointments or other system impacts may preclude traditional physician-led clinics from providing opportunities for in-depth discussions about patients’ and caregivers’ understanding of disease implications or their psychosocial concerns. Since our patient and caregiver

participants seem proactive and engaged, we speculate whether another—perhaps nurse-led model— might be advantageous for collaborating with patients to develop strategies for self-management.

6.6.1 Limitations

This is a small, highly contextualized study and may not reflect other clinical settings. For instance, many DM1 and HD clinics in the United States are multi-disciplinary ‘centers of excellence’ funded by the Muscular Dystrophy Association (MDA, 2014) or the Huntington’s Disease Society of America (HDSA, 2014).

We recruited patients followed in clinics at one academic health science center , therefore we do not represent the voices of patients who chose not to seek follow-up or are followed in other clinical settings. Since we speculate that motivations for attendance change over time and that caregivers take a more active role as the patient deteriorates, it is a challenge to recruit individuals in the later stages of disease. These patients may be institutionalized, lost to follow-up, or difficult to interview because of profound speech or cognitive impairments. We did, however, ask participants to speculate about a time when they might choose not to follow-up. Most participants envisioned that they would continue to attend clinic until they were physically unable to come, or if they believed that nothing more could be done to help them. And finally, while the interviews were conducted by a researcher not involved in patient care, participants may still have provided information that they believed the researcher, or their HCP, wanted to hear. Attempts were made to reassure participants that their responses would not affect their care, but we cannot ensure that participants were not impacted by this concern.

We acknowledge that patients’ have variable disease presentations, and that some patients with significant central nervous system involvement— including memory impairments or problems with executive function—may not have the same degree of interest or ability to participate in their health care. However, we speculate that patients may be judged as a homogenous group rather than as individuals with separate concerns, interests and abilities; consequently, their knowledge and contributions to self-management may be overlooked.

6.6.2 Future Directions

Our findings have raised additional questions that are well-suited to qualitative inquiry. For example, while the purpose of this study was to focus on individuals and their family members who are living with manifest disease; the data suggests that clinic attendance prior to diagnosis is often anxiety-provoking and problematic for patients and their families. Additionally, partners and children of affected parents also access clinic staff to seek information and counseling. This leads to questions about who is considered a ‘patient’ and how resources are allocated. Research exploring clinic access by non-affected individuals has implications for health care delivery and potentially could lead to recommendations for better supportive care for caregivers and those at-risk. Exploring the experiences of patients who attend well-funded and resourced, multi-disciplinary clinics might inform both funding agencies and individual practitioners about clinical care models and guidelines for best practice.

6.7 Conclusion

Our findings suggest that patients and their loved ones living with chronic, progressive and life-limiting neurological illness maintain regular attendance at an outpatient neurology clinic because they perceive that the clinic is a ‘safe’ place where they can seek information, personal connection and explore avenues to advocate for their condition and contribute to scientific advancements. Clinic visits are, therefore, a form of being proactive; that is, patients and caregivers experience a benefit from learning about their condition, educating the medical and general communities about DM1 or HD, helping future generations and seeking social support. While motivations for clinic attendance change over time for patients and caregivers, these factors seem to reduce some of the anxiety of living with a life-limiting condition. However, these findings lead us to question whether the current clinical models in this academic center should shift from a physician-led model to one that supports an increased role for allied health professionals. The study shows that people come to clinic primarily for the safe place it provides, and to escape judgment and isolation. Traditional medical models and traditional physician training are less focused on these supportive goals, suggesting that

perhaps nurses, whose training emphasizes more holistic models of care, could provide productive and cost effective leadership of such clinics. Regardless, despite the lack of treatment options to halt or reverse the disease process, on-going clinic attendance appears to be therapeutic in and of itself, and seems to provide a tangible benefit for patients and loved ones living with DM1 or HD. We anticipate that findings from this research will inform health care professionals about DM1 and HD patients' perspectives about being proactive about their clinical care; in turn, this knowledge may contribute to scholarly conversations that may enhance or change current care practices for these and other chronic disease populations with progressive cognitive and behavioral decline.

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Chapter 7

7 “We Like to Think We’re Making a Difference”: Health Care Providers’ Perspectives About Caring for Individuals with Myotonic Dystrophy and Huntington’s Disease

7.1 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. HCPs at one academic centre in Ontario, Canada were invited to participate in semi-structured interviews; 5 neurologists, 2 specialist physicians, a nurse, two social workers and a physiotherapist consented. Three stages of coding (initial, focused and theoretical) were used to analyze participants’ transcripts, and codes were collapsed into themes and categories.

Results: Three categories including *An Evolving Care Approach*, *Evolving 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. Regardless, 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 described that patients benefited from on-going clinical care provided by proactive clinicians.

Conclusion: Providing care for individuals with DM1 and HD is a balancing act. In particular, 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 about physician-led, patient-centered clinical care models, and we propose that a nurse-led model may be more appropriate for addressing patients' and caregivers' complex biopsychosocial needs.

7.2 Introduction

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 (Jaglal et al, 2014). Moreover, shared decision making is considered the 'pinnacle' of patient-centered care (Barry & Edgman-Levitan, 2012), but researchers and clinicians struggle to define what this means in practice, and whether a patient-centered care approach is always feasible, achievable or desirable (Berwick, 2009). 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 (Wilson, Seymour & Aubeeluck, 2011). However, end of life is only one phase of chronic neurological diseases; therefore, it is necessary to explore the complex and evolving needs of patients over a long disease trajectory that may span years or decades. While there are numerous articles that suggest care guidelines for patients with DM1 or HD (Nance, 2007; Roos, 2010; Sturrock & Leavitt, 2010; Turner & Hilton-Jones, 2008; Gagnon, Mathieu & Noreau 2007)—to our knowledge clinicians' perspectives about providing care for these patients along their disease trajectory has 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-form of muscular dystrophy, is a multi-system 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 (Harper, 2001). There is a well-recognized DM1 personality pattern described in the literature suggesting that patients may have low IQ and apathy (Winblad, Lindberg & Hansen, 2005); in turn, patients may miss clinic appointments and seem indifferent about their health (Meola & Sansone, 2007). Similarly, while HD is characterized by impaired motor function, its psychiatric issues --including depression, apathy, anxiety, obsessions and compulsions, impulsivity, irritability and aggression, and psychosis—are often far more debilitating to patients and their families (Roos, 2010; Sturrock & Leavitt, 2010). Moreover, cognitive impairment variably affects individuals with HD, but when present, results in difficulty with executive function, and problems acquiring, processing and remembering information (Bonelli & Hofmann, 2004). 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 (de Die-Smulders et al., 1998; Mathieu, Allard, Potvin, Prevost, & Begin, 1999). Additionally, DM1 patients may experience sudden death secondary to choking or a cardiac event (de Die-Smulders et al., 1998; Mathieu, Allard, Potvin, Prevost, & Begin, 1999), and the suicide rate in HD is higher than the general population (DiMaio et al, 1993).

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” (Nance, 2007, p. 176). Due to 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 (Chouinard et al., 2009). 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 (Chouinard et al, 2009; Gagnon et al, 2007) and HD (Nance, 2007; Nance, 2012). Chouinard et al (2009) proposed a DM1 Management Model that considers the multi-system nature of the disorder, the propensity for individuals to have limited educational, economic and social opportunities, and the lack of knowledge of non-specialist health care providers (Chouinard et al, 2009). Similarly, Nance (2007) 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 multi-disciplinary 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 (Nance, 2012; Roos, 2010; Sturrock & Leavitt, 2010; Turner & Hilton-Jones, 2008). 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 therefore family members may become instrumental for monitoring an individual’s physical and behavioral changes, and ensuring that he or she is following treatment recommendations (Sturrock & Leavitt, 2010).

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 for patients with other chronic diseases that present with cognitive or behavioral impairments. Therefore, our purpose was to explore how health care professionals perceive of-- and provide care for—individuals with DM1 or HD throughout their disease course. How do they approach the care of these individuals, how effectively do they feel they are meeting patients’ needs, and how does their approach to care evolve over the course of the illness?

7.3 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 was informed by constructivist grounded theory, a qualitative research methodology that studies basic social processes to develop an explanatory theory (Charmaz, 2006; 2014). Patients with mild to moderate DM1 or HD (by physician report), their caregivers, and health care professionals were purposively sampled from the neuromuscular and movement disorders outpatient clinics at an academic medical center in Ontario, Canada. Table 1 provides a snapshot of the clinical contexts. Participants were invited to participate in semi-structured interviews; 14 patients (n= 5 DM1), 10 caregivers (n= 2 DM1), and 11 HCPs including five neurologists, a psychiatrist, a respirologist, a nurse, two social workers, and a physiotherapist consented. We ceased recruitment when we determined that the collected data was 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 health care providers; patient and caregiver data will be reported elsewhere (LaDonna, Watling, Ray & Venance, In Preparation)

Table 7-1: Clinical Care for DM1 and HD Patients at One Academic Center in Ontario, Canada

	Myotonic Dystrophy (DM1)	Huntington's Disease (HD)
Clinical Cohort	<ul style="list-style-type: none"> Approximately 150 patients are followed every 6 months to 2 years. 	<ul style="list-style-type: none"> Approximately 125 patients are followed every 3-6 months.
Clinical Context	<ul style="list-style-type: none"> DM1 patients are seen in a general adult muscle disease clinic. 	<ul style="list-style-type: none"> HD patients may be seen in the monthly HD multi-disciplinary clinic. <p>--Or--</p> <ul style="list-style-type: none"> in a general movement disorders clinic.

Staff/Personnel	<ul style="list-style-type: none"> • 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. 	<ul style="list-style-type: none"> • Four neurologists specialize in movement disorders; one staffs the multi-disciplinary clinic. • A psychiatrist and social worker staff the multi-disciplinary clinic; their services are also available by referral. • There is not a nurse affiliated with the HD multi-disciplinary clinic. • Medical students, residents or fellows may participate in care.
Management	<ul style="list-style-type: none"> • 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. 	<ul style="list-style-type: none"> • Neurological examination, evaluation of motor and psychiatric symptoms, and assessment of psychosocial needs. • Medical treatment: anti-depressants or anti-psychotics 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.

CP conducted all of the HCP interviews, which were recorded and transcribed verbatim; in turn, KAL coded each line or sentence of the first two transcripts from each participant group using words or phrases that captured the experiences or actions described by the participants. Next, KAL consolidated the most frequently occurring codes into preliminary categories, and used these to code the next six transcripts to determine their fit and relevance. KAL met frequently with SLR and SLV to discuss preliminary findings. The research team finalized a list of categories by consensus; KAL then re-coded the entire dataset. Throughout the research process, data within and between transcripts were constantly compared, and KAL 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. This study was approved by the Western University Research Ethics Board; to protect confidentiality, all participants were given a pseudonym.

7.4 Results

Three categories including *An Evolving Care Approach*, *Evolving Roles*, and *Making a Difference* were identified. The participants described that their approach to care evolved along the disease trajectory and 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, while 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.

7.4.1 An Evolving Care Approach

7.4.1.1 First contact

Patients are typically referred to a specialty neurology clinic for three reasons: (1) they are at-risk for inheriting DM1 or HD; (2) they are gene positive but pre-symptomatic; or (3) they have neurological symptoms with or without a family history. Initial visits typically include a review of symptoms and family history, and a comprehensive neurological exam. 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 pre-symptomatic 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).

7.4.1.2 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 60 minute visit for both DM1 and HD consists of a neurological exam 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 anti-depressants 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).

While the clinicians' approaches were similar at both clinics, the structure of care and the role of allied health professional participants differed. While the neuromuscular clinic team has access to allied health professionals including physical therapy and social work, 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, allied health professional). 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 multi-disciplinary 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, allied health professional).

While 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).

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, nurse).

7.4.2 Evolving 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. Moreover, participants recognized the lack of curative treatments for DM1 and HD, and therefore defined their role as primarily one of providing hope by: (a) providing expert evaluation and

education; (b) preventing and managing crises, (c) being an advocate, and (d) providing support. These roles were not seen as mutually exclusive; rather, they were perceived as inter-related 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 they had not been forgotten by the wider medical and research communities:

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 *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).

7.4.2.1 Providing expert evaluation and education

Prior to, 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 8,000, 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 information about funding or support resources. Some HCPs prioritized keeping abreast of the latest research and pharmaceutical options then distilling information for patients. While 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).

7.4.2.2 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 (*DMI*) 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 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 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, allied health professional).

7.4.2.3 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 lynch pin...for linking people to resources" (Dr. Thompson, neurologist). Moreover, 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.

7.4.2.4 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 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 psycho-social 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, allied health professional).

7.4.3 Making a Difference

7.4.3.1 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 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 1½ 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, nurse).

Moreover, 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).

7.4.3.2 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: "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 (*DMI*)” (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).

7.5 Discussion

When asked whether patients benefit from regular on-going follow-up, Martha (nurse) responded:

I’m biased, because I’m the one that’s giving them care. It’s hard to know. I think there, again, 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--because I don’t think that study’s been done. We like to think we’re making a difference, but I don’t think we know that.

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 on-

going 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 (CCM-NC) interviewed 180 HCPs, community members and policy makers identified similar challenges (Jaglal et al, 2014). While 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, however, suggest that patient-centered care provision for patients with DM1 and HD is a balancing act; 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.

Moreover, HCPs must constantly temper their frustrations and sense of futility with the perceived rewards of caring for individuals with DM1 and HD. While 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. Our findings that HCPs have limited time to address caregivers' needs resonate with other providers caring for chronic neurological conditions (Jaglal, 2014; Nance, 2009). In particular, the current physician-led model at our academic centre 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—while 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—was echoed in a small qualitative study examining the perspectives of physicians caring for patients with Alzheimer's disease (Hinton et al., 2007). In essence, there was a sense

amongst our participants that the current physician-led care model at our academic centre 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 with their reliance on—and need to support—caregivers. While 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 (Sturrock & Leavitt, 2010). In addition, our findings resonate with previous literature (Heatwole, 2012; LaDonna, Koopman, Ray & Venance, In Press) that suggests that issues of clinical concern to HCPs may not be what patients and caregivers are aware of or want to address. In particular, while HCPs focus on symptoms that may cause morbidity or mortality, patients and caregivers are generally concerned with issues that impact their relationships (Cup et al, 2011) and participation in education, employment, and leisure and recreational activities (Gagnon, Mathieu & Noreau, 2007). 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 patient-centered clinical care models: Is care still ‘patient-centred’ 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-centred 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 multi-disciplinary 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 multi-disciplinary 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 addressing the evolving needs of patients and caregivers.

Furthermore, 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 more important as patients’ symptoms progress, perhaps because their expertise is better aligned to the needs of patients and families with evolving chronic illnesses. We therefore propose that alternate models of care and leadership, including family health care teams, or rehabilitation or nurse-led 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 (Jaglal, 2014). Moreover, multi-disciplinary, nurse-led clinics are the standard model for other chronic disease populations including heart failure (Strömberg et al, 2003) cancer (Moore et al, 2002), and diabetes (Denver, Barnard, Woolfson & Earle, 2003); this research suggests that patients attending nurse-led clinics have improved self-care behaviors and/or better outcomes. Our findings support

Chouinard and colleagues (2009) who proposed a nurse-led model for the care of patients with DM1. While physicians remain integral to diagnosis and treatment, we propose that nurses are 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 on-going support. Nurses are trained to see--and provide emotional support—to the patient and caregiver as a unit (personal communication, Wilma J. Koopman), and may therefore be best-suited to creating a comfortable clinic space where patients and caregivers can have their complex biopsychosocial needs addressed.

Work is currently being done to create and assess a nurse-led, integrated clinical care pathway for DM1 at a neuromuscular clinic in Quebec (Chouinard et al 2009, Gagnon et al, 2010). The DM1 clinic is led by a nurse care manager (NCM) who works with an interdisciplinary team to fulfill the essential care roles identified by our study participants; that is, the NCM monitors symptoms, treats complications, educates, and supports the psychosocial needs of the patient and his or her family (Gagnon et al 2008). Moreover, several clinics for DM1 and HD in the United States are funded by patient advocacy groups that support a multi-disciplinary group of clinicians to provide care and present research opportunities to patients and families (Muscular Dystrophy Association, 2014; Huntington's Disease Society of America, 2014). However, to our knowledge, while multi-disciplinary, nurse-led or the CCM-NC clinical models show promise, they have not yet been systematically evaluated or implemented across North America (Jaglal, 2014; Gagnon et al 2008).

7.5.1 Limitations and Future Directions

This is a small study describing the perceptions and experiences of health care providers caring for patients with DM1 and HD at one Canadian academic centre. We recognize that clinics and treatment approaches may vary at different locations, and our highly contextualized study is therefore not generalizable to other settings.

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 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. However, since care for patients with DM1 and HD is complex, future research should explore the perspectives of nurses, family physicians and other care allied health professionals. Finally, it is essential to understand the experiences and health care expectations of DM1 and HD individuals and their families prior to proposing a model of care.

7.6 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. Moreover, participants identified a need for greater involvement from allied health professionals, and we suggest that nurses are well-suited 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. Regardless, we propose that research exploring clinical models for patients with complex physical, cognitive 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.

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Chapter 8

8 Discussion

Patient-centered care—“care that is respectful of and responsive to individual patient preferences, needs, and values” (Institute of Medicine, 2014)—may be challenged by DM1-affected individuals’ complex needs (Gagnon et al, 2010; Heatwole et al, 2012), and the lack of disease-halting or curative treatments. Limited research has explored the perspectives of DM1-affected individuals and their caregivers, and researchers have not yet used qualitative methodologies to explore patient-centered care provision in DM1. Similarly, patients’ motivations for maintaining follow-up at a neurology outpatient clinic are unknown. The relative absence of DM1 patients’ experiences in the literature questions whether their voices are being heard or if their psychosocial needs are being met.

Thus, the purpose of this research was to add patients’ and caregivers’ perspectives to the literature and to explore how patient-centered care provision for DM1 is perceived—and if it is feasible—at one academic centre in Ontario, Canada. Three qualitative methodologies were used to illuminate patients’ and caregivers’ experiences living with DM1 (Chapters III and IV), their health care expectations, and their motivations for clinic attendance (Chapter VI). Individuals living with Huntington’s disease and their caregivers were purposively sampled to increase the breadth and depth of the experiences of clinical care for patients with inherited and life-limiting neurological conditions (Chapter VI). I also explored health care providers’ perspectives about providing care for these patients and families (Chapters VI; Chapter VII). Findings from this research suggest that there are numerous challenges—and rewards—to effective patient-centered care provision for DM1 and HD. However, most patient, caregiver and HCP participants perceived that affected individuals and their families receive tangible benefits from maintaining follow-up at a neurology outpatient clinic. Patient and caregiver participants described being proactive seekers of information and care, and they conceptualized the

neuromuscular and movement disorders clinics at our institution as a ‘safe place’ (Figure 8-1).

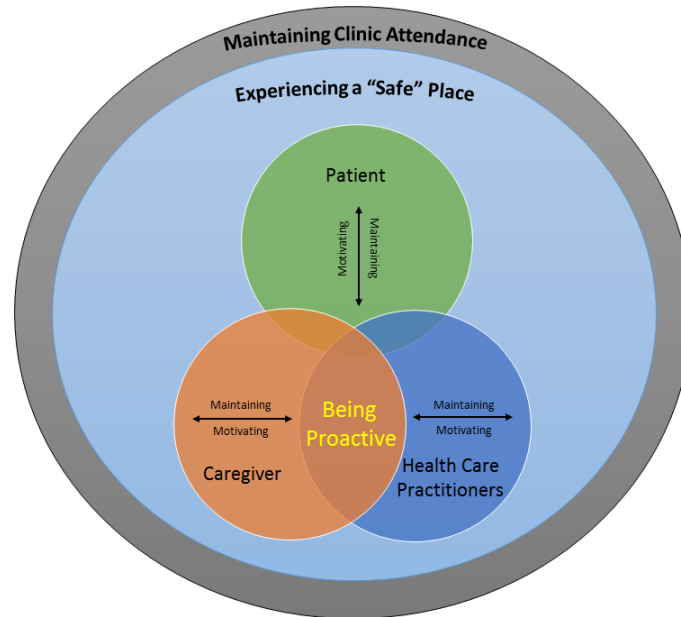


Figure 8-1: Clinic is a “Safe Place”

8.1 What is Already Known

8.1.1 Myotonic Dystrophy (DM1)

DM1 is a multi-system disorder that is characterized by an unrelentingly progressive and devastating disease course. The muscular, cardiac, respiratory, endocrine, gastrointestinal and central nervous systems are affected, and primary clinical manifestations include progressive muscle weakness, muscle stiffness due to delayed relaxation (myotonia), arrhythmias, excessive sleepiness, early-onset cataracts, and cognitive and behavioral manifestations, namely low IQ and apathy (Harper, 2001). In particular, “the general clinical impression in most cases is one of apathy, decreased emotional participation, and psychomotor delay” (Meola & Sansone, 2007, p. 296), and there is a well-recognized DM1 personality pattern described in the literature (Winblad, Lindberg & Hansen, 2005). In particular, “a TCI (Temperament and Character Inventory) based portrait of the average DM1 patient, then, is that of a private, introverted individual

with little self-esteem, burdened by fatigue and low energy” (Winblad, Lindberg & Hansen, 2005, p. 290). While it is unknown if the emotional symptoms are a reaction to the disease process or an organic part of the disorder caused by central nervous system lesions (or both), it is important for health care providers to assess cognitive and behavioral impairments because they may have a significant effect on an individual’s quality of life (Bungener, Jouvent & Delaporte, 1998).

Qualitative and quantitative inquiries have described the impact of DM1-affected individuals’ physical, cognitive and emotional symptoms on social participation (Gagnon, Mathieu & Noreau, 2007; Gagnon et al, 2008), their relationships (Cup et al, 2011), and their quality of life (Laberge et al, 2013)). Care for DM1-affected individuals may be challenged by their variable symptom presentations (Heatwole, 2012; Gagnon et al, 2010) lifestyle risk factors and challenges engaging in health promotion behaviors (Gagnon et al, 2013). Apathy and disinterest in their health have been attributed causality for missed clinical appointments (Meola & Sansone, 2007) or failure to adhere to treatment recommendations (Chouinard et al, 2009). Guidelines for DM1 care have been proposed (Gagnon et al 2007, Chouinard et al, 2009), but these have not yet been systematically implemented or evaluated. Potentially, one of the main challenges facing the implementation and evaluation of patient-centered care provision is the lack of a uniform conceptualization or definition of what ‘patient-centered’ care is and how it is applied in practice. In particular, clinicians and researchers debate the feasibility, achievability and desirability of taking a patient-centered care approach (Berwick, 2009).

8.2 What Participants Have Taught Us

8.2.1 Challenges to providing patient-centered care for individuals with DM1

While participants described that on-going clinic attendance is beneficial for DM1-affected individuals and their caregivers, this research uncovered several challenges to patient-centered care.

8.2.1.1 Variable knowledge

Studies describe that DM1 patients' have variable understanding about their condition (Laberge et al, 2010; Faulkner, 1998) and I speculate that this impacts clinical practice. Our findings suggest that while some participants were well-informed about their condition and were able to articulate their symptoms, some individuals accepted misinformation about DM1 as fact—particularly in regard to genetic information—that they in turn used to make important decisions (Chapter V). This occurred despite formal genetic counseling consultations and discussions during clinic visits. Previous research suggests that individuals with DM1 either do not accept their diagnosis or do not understand the hereditary ramifications of their condition (Nätterlund, Sjöden & Ahlström, 2001). These findings resonate with the present research, but we suggest that poor understanding of chronic health conditions is not specific to DM1; in particular, patients with other chronic health conditions may also have difficulty synthesizing and understanding complex genetic information (McKibbin et al, 2014). It may therefore be necessary to assess patients' knowledge at each clinic visit, or to provide patients with more frequent follow-up—perhaps with a nurse or social worker—to provide ample opportunity for patients to ask questions and to resolve knowledge gaps.

8.2.1.2 Striking a Balance: Providing Care for Patients with Chronic Neurological Conditions

Findings from *Hard to Swallow* (Chapter III) and *Picturing the Experience* (Chapter IV) resonate with previous literature suggesting that patients and clinicians may have divergent clinical care goals (Heatwole et al, 2012). While speculative, it appears that patients and caregivers focus primarily on symptoms that impact their daily activities and quality of life (Chapter III, Chapter IV), while clinicians focus on symptoms that either have the potential to cause serious complications or result in sudden death, and/or those that they are able to treat. Some of the caregivers in the *Hard to Swallow* study were perplexed that I was interested in studying dysphagia since they perceived that their loved one had to face far more troubling symptoms like weakness and fatigue. Similarly, none of the patient participants in the *Picturing the Experience* study volunteered swallowing

dysfunction as one of their troubling symptoms, and only reflected on it when asked direct questions about their diet. It is possible that dysphagia develops slowly and individuals are therefore able to compensate for their swallowing problems by developing strategies like avoiding certain foods, regurgitating objects or having liquids with meals. We speculate that caregivers' lack of concern mirrors their loved one's attitudes about swallowing dysfunction; that is, swallowing is—in essence—not on patients' or caregivers' 'radar' because patients have found ways to cope with, or adapt to this slowly progressive symptom. In contrast, weakness and fatigue significantly impacted participants' ability to work or participate in recreational or leisure activities. This finding resonates with previous research that describes symptom impact on DM1-affected individuals' social participation (Gagnon, Mathieu & Noreau 2007; Gagnon et al, 2008) and health related quality of life (Antonini et al, 2006; Peric et al, 2010; Laberge et al 2013). Cup et al (2011) also described the impact of chronic neuromuscular conditions on the marital relationship and on caregivers' quality of life (Geirdal, Lund-Petersen & Heiberg, 2014). Our caregiver participants' described having to prod loved ones to leave the house, exercise or participate in social activities. Some caregivers also described that their loved ones' fatigue impacted their relationship; that is, individuals with DM1 were either too tired to participate in activities, or they would fall asleep at inappropriate times – sometimes in the middle of a conversation.

We speculate that physician-led clinical care models typically address diagnosis and treatment; consequently patients' and caregivers' educational and psychosocial needs may not be sufficiently managed. HCP participants described that allied health professionals should become more involved in care as patients' symptoms progress (Chapter VII); we therefore suggest that there is a need for a holistic clinical approach that addresses patients' symptoms and emotional needs while supporting patients and families with their daily challenges.

8.2.1.3 Adapting the care plan across the disease trajectory

Findings suggest that clinical care evolves throughout the patients' disease trajectory (Chapters VI and VII). This may occur not only because of progressive functional

decline, but also because of the emotional difficulty patients may experience as they adjust to their diagnosis and their shifting identity (Chapter IV; Nätterlund, Sjöden & Ahlström, 2001). Patients, caregivers and HCPs described that patients' and caregivers' motivations for clinic attendance changed over time. Moreover, while HCPs described having a 'template' for clinic visits, it was evident that they adapted their approach based on a patient's stage of disease and his or her cognitive or behavioral abilities.

Despite the lack of disease-halting or curative treatments— and other HCP-identified challenges providing patient-centered care for individuals with DM1 (and HD)— all participants described that patients and caregivers experience tangible benefits from on-going clinical care. Findings from an exploratory study in the United Kingdom that examined the experiences of patients attending a neuromuscular disease rehabilitation centre found that patients attend clinic to obtain knowledge and understanding about their condition from experts, and to receive community support from individuals with similar conditions (Hartley, Goodwin & Goldbart, 2011). In turn, the rehabilitation centre may become a place of “empathy” that can provide hope for patients (Hartley, Goodwin & Goldbart, 2011).

However, ‘hope’ was not universally experienced by patients; instead, patient and caregiver participants seemed to attend clinic to be understood, and to collaborate with experts to educate the general and medical communities. Participants were aware that there was likely little that could be done to help them; consequently, participants wanted to help future generations by collaborating with experts to work toward therapeutic advancements and ‘fight’ for a cure. This resonates with the experiences of patients with other chronic illness (Karlawish, Casarett & James, 2002; Tong et al, 2008 , Madsen, Holm & Riis, 1999) but was perhaps an unexpected finding given the literature that DM1 patients may lack awareness or motivation (Chouinard et al, 2009; Meola & Sansone, 2007).

The idea of clinic as a place of hope may therefore need to be re-considered; instead, we suggest that clinic be conceptualized as a place to empower patients through education and collaboration. For example, patients and their families may not fully appreciate the

implications of their life-limiting symptoms, so clinic is an ideal—and perhaps safe—place for experts to educate patients and their families, and engage them in conversations about self-management. Clinic visits are also an opportunity for HCPs to collaborate with patients to advocate for research or to educate medical and other health professional students and the general public about these uncommon illnesses.

8.2.2 Being Proactive

Despite challenges, the patients at the centre of the care at our institution demonstrated that they actively seek information about their condition and opportunities to make a difference for future generations. This research found that, in general, the patient participants attended their clinic appointments and were interested in their health.

Most participants in the studies described in Chapters III-VII were actively engaged and articulate about the impact of symptoms on their daily lives and their health care expectations. I recognize that individuals with DM1 have variable disease presentations, and that our research participants were purposively recruited. However, we speculate that—in general—DM1-affected individuals' physical appearance and speech impairments may cause them to be unfairly labeled by the general public or health care providers. Participants in this research perceived that they were judged by members of their community. This resonates with previous muscular dystrophy literature that suggests that individuals living with different forms of muscular dystrophy experience judgment, shame or stigma (Boström & Ahlström, 2004). However, the photovoice participants described that they found solutions for coping with their shifting identity and for managing the challenges of everyday activities. Participants were resilient, and problem-solved solutions for daily challenges like opening doors, jars or bottles, and participants were also able to mitigate difficulties with study procedures. For instance, participants recruited friends or caregivers to assist them with picture taking, participate in their individual interview, or transport them to the clinic visit.

Patients were not only active research participants, but also active in deciding if and when to seek health information and social support (Chapter VI). Some chose not to attend support groups or to read about DM1, while others actively sought information

from medical journals and patient advocacy groups (Chapters III-VI). Participants took their role as patient educators seriously; that is, participants stated that their clinic attendance taught HCPs about their uncommon, variable and unpredictable condition (Chapter VI). A qualitative analysis of interviews and focus groups conducted with 30 patient educators/mentors living with arthritis found that sharing their stories with health professional students helped them “reframe” their illness from a negative experience into something that makes a positive contribution to student learning. In turn, the patient educators believed that their contributions may improve the wider health care system (Laukner, Doucet & Wells, 2012). Similarly, our participants valued their own expertise, and took it upon themselves to educate non-specialist clinicians or members of the public by giving them information about DM1 or participating in patient advocacy groups (Chapters III-VI). One of our study participants is a leader in the DM1 patient advocacy community, and another shared his pictures and experiences with a graduate class in occupational therapy to teach future clinicians about DM1.

8.3 What This Research Adds

This research is the first to explore caregivers for individuals with DM1 and dysphagia, collaborate with participants using photovoice, and include the perspectives of patients, caregivers, and HCPs about the expectations and motivations for ongoing clinic attendance. While the experiences of caregivers for individuals with muscular dystrophy have been explored, these studies did not specifically address the experience of providing care for an individual with dysphagia. Moreover, caregiving for DM1-affected individuals has not previously been explored in the context of caregivers’ lived time, space, bodies and relationships. This work contributes to a deeper understanding of the perspectives of providing care for individuals with DM1, and supports previous research suggesting that symptoms that clinicians prioritize are not necessarily those that are most concerning for individuals with DM1 and their caregivers (Heatwole et al, 2012).

This research used photovoice—a novel and innovative qualitative methodology—to engage DM1-affected individuals as research collaborators. The visual representation of patients’ perspectives of their illness experience—including their knowledge and

misperceptions about DM1 (Chapter IV)—is a novel contribution to the literature. This research also contributes photographic representations of DM1 patients’ problem-solving techniques and provides insight into how they cope with the challenges of daily activities.

Previous research has explored European neuromuscular disease patients’ experiences of attending a rehabilitation centre (Hartley, Goodwin & Goldbart, 2011); however, our research advances this work by contributing an exploration of patients’, caregivers’ and health care providers’ perceptions of health expectations and motivations for on-going clinic attendance across the disease trajectory. To our knowledge, DM1 and HD have not previously been explored together to provide a greater sense of the experiences of patients living with genetic, life-limiting neurological conditions. The idea that patients’ and caregivers’ motivations for clinic attendance change over time has also not been reported. From this research, we learned that DM1 and HD individuals and their caregivers are proactive about managing patients’ health, and are driven by symptomatic management, altruism, and reassurance. In particular, they look for information and social support in a clinical context that they experience as ‘safe’. Previous work has described a neuromuscular clinic as a place of “empathy” (Hartley, Goodwin & Goldbart, 2011) that provides supportive care (Gagnon et al, 2010). However, DM1 and HD patients’ and caregivers’ conceptualization of clinic as a ‘safe place’ is a novel finding. This research contributes that—in the absence of disease-halting or curative pharmaceutical therapies—clinic attendance for these individuals is considered treatment.

To our knowledge, neither HCPs descriptions of their roles caring for patients with DM1 and HD, nor the challenges and rewards of patient-centered care for this population, have previously been reported. HCPs describe patient-centered care for DM1 and HD as a ‘balancing act’ in which HCPs have to balance (1) patients’ medical and psychosocial needs, (2) their sense of futility that their best efforts will not prevent patients from deteriorating, (3) their rewards for caring for these individuals (4) and the need to be a patient advocate while addressing caregivers’ concerns. Hope might be expected as a dominant theme and has been suggested as a reason for creating neuromuscular disease clinics (Hartley, Goodwin & Goldbart, 2011), and here, HCPs identified providing hope as one of their main roles in the care of these individuals. However, hope was not

universally experienced—or sought—by patients and their caregivers. Instead, participants appeared more interested in proactively seeking reassurance that they were being cared for, and that they were collaborating with HCPs to find answers to their uncommon conditions. Our novel research findings suggest that maintaining clinical follow-up empowered participants to exert control over their unpredictable illness. Clinicians and researchers may therefore need to rethink assumptions about what patients may want from their care, and perhaps offer patients opportunities—including research, education or advocacy roles—that will enable them to make tangible contributions to their health. Finally, perhaps the most novel and significant contribution of this work is that it has identified that individuals with DM1 are proactive research participants and collaborators, advocates and educators.

8.3.1 Implications

Findings from this research have started to change practice. One clinician noted that seeing the photographs and reading participants' experiences from the *Picturing the Experience* (Chapter IV) study was the first time that she felt that she understood and 'knew' her DM1 patients. Prior to reading the manuscript, this clinician had difficulty identifying whether patients were interested in participating in clinic or whether they fully grasped the implications of DM1. However, the photovoice study gave this clinician a tangible 'picture' of patients' lives, and she was therefore better able to understand how patients cope with their condition.

Another clinician stated that she altered her approach to clinic visits after reading the *Hard to Swallow* manuscript. Previously, this clinician had devoted a significant amount of clinic time to addressing dysphagia at each visit and discussed the importance of diet and swallowing studies. After reading the paper and discovering that swallowing function may not be what patients and clinicians want to spend clinic time addressing, this HCP altered the approach and switched the focus from discussing potential complications to ensuring that patients knew how to safely manage a choking episode (i.e. "what is the universal sign for choking?" "do you know the Heimlich maneuver?").

8.3.1.1 Considerations Using Qualitative Research for Exploring DM1

Relatively few studies have qualitatively explored individuals living with myotonic dystrophy, and it is unknown if patients' cognitive and behavioral impairments have impacted the lack of patient-centered research in this population. However, interviews with DM1 individuals were excluded from previous research because patients' speech impairments complicated interviews, and the researchers' perceived that were not able to reflect on their condition (Nätterlund, Sjöden & Ahlström, 2001). There were challenges using these methodologies in DM1 including (1) difficulty conducting and transcribing interviews with patients' with speech impairments, (2) fatigue precluding participation, and (3) participants' challenges with problem-solving. For example, it was difficult to conduct and transcribe an interview for a participant with dysarthria, one participant withdrew from photovoice because of fatigue, and two participants were unable to use the digital cameras. However, the benefits of using qualitative methodologies and methods with DM1-affected individuals were considerable. All participants—regardless of their symptom presentation and challenges complying with study procedures—had keen insights into their condition and the impact of symptoms on their daily lives. They also were able to articulate their strategies for proactively managing their condition and discussing their health care expectations. I am confident that qualitative research methods and methodologies are appropriate and useful for adding DM1 patients' voices to the literature. All participants were enthusiastically engaged in the research process, and some appreciated having the opportunity to share their stories and educate researchers and clinicians about DM1:

It (*research study*) gives them an idea of what it's like and learn that it's not all negative and that ... like I say, you don't know what it's like unless you live it, but to take pictures you can see what my life is like. Not all negative, but not all positive either. Yeah, it's a good idea (Photovoice Participant 4).

The greatest challenge of using qualitative research came not from patient participants, but from trying to have their voices included alongside those of basic scientists and

clinicians in academic medical journals. Previous research has explored the limited proliferation of qualitative research in these journals (Gagliardi & Dobrow, 2011; Shuval et al, 2011). Two medical journals declined to send our photovoice article for peer review; one editor stated that their journal “did not publish this kind of work” and another sent a thoughtful email in which he expressed his personal interest in our manuscript but stated that financial limitations and page restrictions prevented him from publishing “sociological” work. Given that ‘patient-centered’ care is a ‘hot topic’ in medical education and care provision, it is puzzling that medical journals are reluctant to publish qualitative work that directly reflects patients’ and caregivers’ experiences. However, we suspect that this is not an uncommon experience despite calls for greater inclusion of qualitative work in medical journals (Malterud, 2001), and discussions about the importance and usefulness of qualitative methodologies for exploring neurological conditions (Macdonald & Chalk, 2011). I have argued throughout this research that patient-centered care cannot be examined without exploring patients’ experiences and needs. Qualitative research is well-suited to this, and steps—including educating the wider medical community about the value and rigor of qualitative inquiry, finding reviewers skilled in qualitative research for scientific journals, and creating better funding opportunities for qualitative research—need to be implemented in order for qualitative research to become more widely-distributed (Krumholz, Bradley & Curry, 2013).

8.3.1.2 Future Directions

The theme of ‘patients (and caregivers) as educators’ was identified across my research studies, and this is an important area for future exploration, particularly since anecdotal evidence suggests that two clinicians changed aspects of their practice based on patients’ and caregivers’ perspectives. A modified grounded theory analysis of 356 second year medical students’ written reflections about the role of patient educators in their undergraduate medical education found that patient educators enabled students to see chronic disease in context with patients’ lives. In turn, they were better able to understand the complexities of patient-centered care. Students viewed patient educators as experts and collaborators who enhanced their medical education (Oswald, Czupryn, Wiseman & Snell, 2014). Therefore, I suggest that patients should be encouraged to

speak to medical and other health professional students, and participate in medical rounds presentations with consultant specialists. In particular, future studies might pair medical students with patients to conduct or evaluate research.

However, while some DM1 participants described themselves as educators who were responsible for teaching the general public or non-specialist HCPs about their uncommon condition (Chapter IV; Chapter VI), they had variable understanding about their DM1 (Chapter V). We identified knowledge gaps—particularly in regard to genetic information—but we did not formally assess participants’ health literacy. Since poor health literacy can negatively affect patients’ health outcomes (DeWalt et al, 2004), a formal assessment of health literacy in DM1 is warranted.

Similarly, patients and caregivers may lack awareness about the implications of their symptoms; consequently, patients’, caregivers’ and HCPs’ goals may not align. This raises important questions about how clinicians can address issues that may contribute to morbidity and mortality while addressing concerns that are important to patients and their caregivers. I suggest that the first step is to continue to ask patients what is important to them and to disseminate this information to clinicians and researchers; in turn, additional research and better education for patients, caregivers and clinicians is necessary to highlight the issues of concern and develop innovative ways to create patient education materials or other educational initiatives. For example, including participants’ photographs in educational materials may be a novel way to share coping strategies with the DM1-patient community. Further, nurses and allied health professionals may close some of the gaps by addressing psychosocial concerns while physicians provide medical surveillance.

This research suggests that patient-centered care in DM1 warrants further investigation and focus the role of caregivers in clinical care. It may therefore be important to re-frame the conceptualization of ‘patient-centered’ care to one that is ‘patient and family centered’. In particular, I question whether the physician-led clinical care model at our academic centre is the most efficient approach for meeting patient and caregiver participants’ complex needs. Findings suggest that physicians may provide care on the

‘margins’ of the illness experience; that is, they are able to provide education and symptomatic management, but HCP participants identified that they have limited time—and perhaps training—to address psychosocial needs. While physicians are integral to DM1 patient care, I speculate that consolidated clinic models—such as family health care teams, rehabilitation centres or nurse-led clinics—may be important to explore. An integrated care pathway led by a nurse care manager (NCM) in Quebec has been evaluated, and findings suggest that the NCM provides systematic, comprehensive and evidenced based follow-up to DM1-affected individuals. This evaluation of the NCM is a first step to developing the role of nursing at other neuromuscular clinics, but has not yet been formally implemented or evaluated at other centres (Gagnon et al, 2010).

8.3.2 Trustworthiness

A number of strategies can be used by qualitative researchers to establish the trustworthiness of their findings. Guba & Lincoln (1985) suggest that trustworthy qualitative research demonstrates credibility, transferability, dependability and confirmability. We used several of the approaches described by Shenton (2004) to ensure that these criteria were met. Member checking, an iterative data collection and analysis process, and peer debriefing and review were used to ensure that we represented a “true picture” (Shenton, 2004, p. 63) of participants’ experiences (credibility). Member checking was part of the iterative data collection and analysis process; that is, questions were added to the semi-structured interview guides to probe themes identified in early interviews (Chapter III, IV and VI). We were therefore able to determine whether experiences resonated with multiple participants; in addition, we purposively recruited two participants for the photovoice study (Chapter IV) to add their perspectives and to review preliminary study findings with them. Additionally, the research team met frequently to discuss the analysis; any discrepancies were resolved by consensus. Finally three of the studies have been peer reviewed (Chapter III; Chapter VI; and Chapter V); one has been accepted (Chapter III), one accepted with revisions (Chapter IV), and one invited for further consideration (Chapter V). Transferability—rather than generalizability—is the goal of qualitative inquiry. We ensured that readers have enough information to evaluate resonance and whether findings can be applied to another setting

by providing a detailed description of the research context and participants. In addition, there is an extensive audit trail of each study that details the data collection and analysis procedures; other researchers can therefore recreate our studies (dependability), although we anticipate that the co-construction of findings from different researchers and participants might produce different interpretations. Finally, a combination of previously mentioned strategies—including a detailed audit trail, member checking and iterative data collection and analysis—ensures that our findings were identified through rigorous and emergent research (confirmability).

8.4 Conclusion

This research sought to explore the experience of living with—and providing care for—DM1; three qualitative research methodologies were used to examine questions pertaining to lived experience and patient-centered care provision. The objectives of this research (1) to add patients and caregivers voices to the literature and (2) explore health care expectations and motivations for on-going clinic attendance to add to scholarly conversations about patient-centered care provision in DM1 have been met. Patients and caregiver participants were proactive about seeking health information, managing their care, and were engaged research collaborators. While care for DM1 patients is complicated by their variable symptom presentations, the lack of disease-halting or curative treatments, and limited time to address patients' and caregivers' psychosocial needs, findings suggest that patients and caregivers benefit from regular clinical care. Findings have important implications for patient-centered care provision in DM1; that is, we have identified that current physician-led clinical models at our academic centre may not be meeting the complex biopsychosocial needs of individuals with DM1, and we suggest future avenues for research. Moreover, the variable definitions and conceptualizations of patient-centered care make it difficult—if not impossible—to evaluate current models. Therefore, a unified patient-centered care definition—based on the experiences of patients, caregivers, and clinicians—needs to be devised, implemented and evaluated; qualitative research is well-suited to this inquiry.

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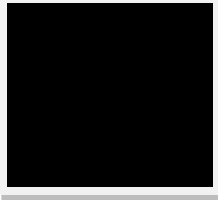
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Appendices

Appendix A: REB #15559E A Study of the Lived Experiences of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia - Original Approval



Use of Human Participants - Ethics Approval Notice



Principal Investigator: [Redacted]
Review Number: 15559E
Review Level: Delegated
Approved Local Adult Participants: 10
Approved Local Minor Participants: 0
Protocol Title: A Study of the Lived Experience of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia
Department & Institution: Clinical Neurological Sciences, University of Western Ontario
Sponsor:
Ethics Approval Date: July 28, 2011 **Expiry Date:** October 31, 2012
Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Study End Date	The study end date has been revised to October 31, 2012 to allow for project completion.	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the UWO Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The UWO HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.

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Appendix B: REB #15559E A Study of the Lived Experiences of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia - July 2011 Revision



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 Review Number: 15559E
 Review Level: Delegated
 Approved Local Adult Participants: 10
 Approved Local Minor Participants: 0
 Protocol Title: A Study of the Lived Experience of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia
 Department & Institution: Clinical Neurological Sciences, University of Western Ontario
 Sponsor:
 Ethics Approval Date: July 28, 2011 Expiry Date: October 31, 2012
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Study End Date	The study end date has been revised to October 31, 2012 to allow for project completion.	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the UWO Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

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Appendix C: REB #15559E A Study of the Lived Experiences of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia - October 2012 Revision



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 File Number: 101015
 Review Level: Delegated
 Approved Local Adult Participants: 10
 Approved Local Minor Participants: 0
 Protocol Title: A Study of the Lived Experience of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia - 15559E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: October 12, 2012 Expiry Date: October 31, 2014
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Study End Date	The study end date has been revised to October 31, 2014 to account for the leave of a team member.	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/CH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the University of Western Ontario Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.



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Appendix D: REB 17995E - Picturing the Experience of Living with DM1



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 Review Number: 17995E
 Review Level: Delegated
 Approved Local Adult Participants: 10
 Approved Local Minor Participants: 0
 Protocol Title: Picturing the Experience of Living with Myotonic Dystrophy (DM1)
 Department & Institution: Clinical Neurological Sciences, University of Western Ontario
 Sponsor:
 Ethics Approval Date: May 30, 2011 Expiry Date: April 30, 2013
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
UWO Protocol		
Letter of Information & Consent		2011/04/12
Letter of Information & Consent		

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The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the UWO Updated Approval Request Form.

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The Chair of the HSREB is Dr. Joseph Gilbert. The UWO HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.

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Appendix E: REB 17995E - Picturing the Experience of Living with DM1



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 Review Number: 17995E
 Review Level: Delegated
 Approved Local Adult Participants: 10
 Approved Local Minor Participants: 0
 Protocol Title: Picturing the Experience of Living with Myotonic Dystrophy (DM1)
 Department & Institution: Clinical Neurological Sciences, University of Western Ontario
 Sponsor:
 Ethics Approval Date: May 30, 2011 Expiry Date: April 30, 2013
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
UWO Protocol		
Letter of Information & Consent		2011/08/18
Letter of Information & Consent		

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the UWO Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The UWO HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.

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Appendix F: REB 17995E - Picturing the Experience of Living with DM1 - February 2013 Revision



**Western
Research**

Use of Human Participants - Ethics Approval Notice

Re:

Principal Investigator: [REDACTED]
 File Number: 100308
 Review Level: Delegated
 Approved Local Adult Participants: 10
 Approved Local Minor Participants: 0
 Protocol Title: Picturing the Experience of Living with Myotonic Dystrophy (DM1) - 17995E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: February 07, 2013 Expiry Date: April 30, 2013
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Western University Protocol	This purpose of this revision is to add a chart review to this protocol. The purpose of the chart review is to ascertain information about the participants' history, symptoms, and topics that the participants and their health care professional(s) deem important for their health. We are also adding Dr. Amer Ghavanini to the list of research personnel. Dr. Ghavanini will be reviewing charts and helping with data analysis. The UWO protocol submission form and the LOI have been revised. Dr. Venance, or another member of the study team, will explain the changes to enrolled study participants, and if they consent, they will be asked to sign a revised consent form.	
Revised Letter of Information & Consent		2013/01/04
Letter of Information	Letter to the participants asking permission to review their chart.	
Recruitment Items	Script for telephone followup to verify that participants received the revised LOI.	
Instruments	Chart Abstraction form (DOB deleted - paper copy)	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the University of Western Ontario Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.

Ethics Officer to Contact for Further Information

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Western University, Research, Support Services Bldg., Rm. 5150
 London, ON, Canada N6A 3K7 t. 519.661.3036 f. 519.850.2466 www.uwo.ca/research/services/ethics

Appendix G: REB 17995E - Picturing the Experience of Living with DM1 - Octoebr 2013 Revision



**Western
Research**

Use of Human Participants - Revision Ethics Approval Notice

Principal Investigator: [REDACTED]
 File Number: 100308
 Review Level: Delegated
 Protocol Title: Picturing the Experience of Living with Myotonic Dystrophy (DM1) - 17995E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: October 28, 2013 Expiry Date: April 30, 2014
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Study End Date		

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the University of Western Ontario Updated Approval Request Form.

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The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.



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Appendix H: REB# 18277E Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - Original Approval



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
Review Number: 18277
Review Level: Delegated
Approved Local Adult Participants: 30
Approved Local Minor Participants: 0
Protocol Title: Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic
Department & Institution: Clinical Neurological Sciences, University of Western Ontario
Sponsor:
Ethics Approval Date: [REDACTED] **Expiry Date:** January 31, 2013
Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
UWO Protocol		
Letter of Information & Consent	Health Care Provider	2011/08/09
Letter of Information & Consent	Patients	2011/08/09

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the UWO Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The UWO HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.



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 Office of Research Ethics
 Support Services Building Room 5150 • London, Ontario • CANADA - N6G 1G9
 PH: 519-661-3036 • F: 519-850-2466 • ethics@uwo.ca • www.uwo.ca/research/ethics

Appendix I: REB# 18277E Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - January 2012 Revision



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 Review Number: 182
 Review Level: Delegated
 Approved Local Adult Participants: 50
 Approved Local Minor Participants: 0
 Protocol Title: Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic
 Department & Institution: Clinical Neurological Sciences, University of Western Ontario
 Sponsor:
 Ethics Approval Date: January 30, 2012 Expiry Date: January 31, 2014
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised UWO Protocol	The variety of health care providers that may be recruited has been expanded, thus increasing the number of Health Care Provider participants to 20.	
Change in Study Personnel	Christine Piechowicz has been added as a research assistant.	
Revised Study End Date	The study end date has been revised to January 31, 2014.	
Revised Letter of Information & Consent		2012/01/16
Revised Letter of Information & Consent	Health Care Provider DM1	2012/01/16
Revised Letter of Information & Consent	Individual with DM1	2012/01/16
Revised Letter of Information & Consent	Individuals with HD	2012/01/16
Letter of Information		2012/01/12
Letter of Information		2012/01/16

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices; Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

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Appendix J: REB# 18277E Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - May 2012



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 File Number: 101031
 Review Level: Delegated
 Approved Local Adult Participants: 70
 Approved Local Minor Participants: 0
 Protocol Title: Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - 18277E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: May 18, 2012 Expiry Date: January 31, 2014
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Caregiver Letter of Information & Consent	DM1	2012/04/10
Caregiver Letter of Information & Consent	HD	2012/04/10
Revised Western University Protocol	A caregiver letter has been added so that caregivers may also participate in the study. The number of participants has been increased to 70.	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the University of Western Ontario Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.

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Appendix K: REB# 18277E Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - June 2012 Revision



Use of Human Participants - Ethics Approval Notice

Principal Investigator: [REDACTED]
 File Number: 101031
 Review Level: Delegated
 Approved Local Adult Participants: 70
 Approved Local Minor Participants: 0
 Protocol Title: Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - 18277E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: June 20, 2012 Expiry Date: January 31, 2014
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Letter of Information & Consent	Individuals with HD - The letter of information has been revised administratively.	2012/06/13

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the University of Western Ontario Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services.



Ethics Officer to Contact for Further Information



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Appendix L: REB# 18277E Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - August 2012 Revision



Use of Human Participants - Ethics Approval Notice

Research Ethics

Principal Investigator: [REDACTED]
 File Number: 101031
 Review Level: Delegated
 Approved Local Adult Participants: 70
 Approved Local Minor Participants: 0
 Protocol Title: Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - 18277E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: August 28, 2012 Expiry Date: January 31, 2015
 Documents Reviewed & Approved & Documents Received for Information:

Document Name	Comments	Version Date
Revised Study End Date	The study end date has been extended to January 31, 2015 to allow for continuation of the project.	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

The ethics approval for this study shall remain valid until the expiry date noted above assuming timely and acceptable responses to the HSREB's periodic requests for surveillance and monitoring information. If you require an updated approval notice prior to that time you must request it using the University of Western Ontario Updated Approval Request Form.

Members of the HSREB who are named as investigators in research studies, or declare a conflict of interest, do not participate in discussion related to, nor vote on, such studies when they are presented to the HSREB.

The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.

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Appendix M: REB# 18277E Understanding the Decision-Making Process for Adults with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic - October 2012 Revision



Use of Human Participants - Ethics Approval Notice

Research Ethics

Principal Investigator: [REDACTED]
 File Number: 101015
 Review Level: Delegated
 Approved Local Adult Participants: 10
 Approved Local Minor Participants: 0
 Protocol Title: A Study of the Lived Experience of Caregivers for Individuals with Myotonic Dystrophy and Dysphagia -15559E
 Department & Institution: Schulich School of Medicine and Dentistry/Clinical Neurological Sciences, Western University
 Sponsor:
 Ethics Approval Date: October 12, 2012 Expiry Date: October 31, 2014
 Documents Reviewed & Approved & Documents Received for Information:

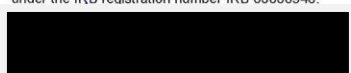
Document Name	Comments	Version Date
Revised Study End Date	The study end date has been revised to October 31, 2014 to account for the leave of a team member.	

This is to notify you that The University of Western Ontario Research Ethics Board for Health Sciences Research Involving Human Subjects (HSREB) which is organized and operates according to the Tri-Council Policy Statement: Ethical Conduct of Research Involving Humans and the Health Canada/ICH Good Clinical Practice Practices: Consolidated Guidelines; and the applicable laws and regulations of Ontario has reviewed and granted approval to the above referenced revision(s) or amendment(s) on the approval date noted above. The membership of this REB also complies with the membership requirements for REB's as defined in Division 5 of the Food and Drug Regulations.

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The Chair of the HSREB is Dr. Joseph Gilbert. The HSREB is registered with the U.S. Department of Health & Human Services under the IRB registration number IRB 00000940.



Website: [REDACTED] to Contact for Further Information



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 t. 519.661.3036 f. 519.850.2466 www.uwo.ca/research/ethics

Appendix N: Dysphagia Study Letter of Information



LETTER OF INFORMATION

PROTOCOL TITLE: A Pilot Study Assessing the Needs of Caregivers for Individuals with Myotonic Dystrophy (DM1) and Dysphagia

INVESTIGATORS: Kori A. LaDonna, BA, PhD Candidate
Shannon L. Venance, MD, PhD, FRCPC

PURPOSE OF THE STUDY: To determine and understand the needs of caregivers who help manage the care of an individual who has been diagnosed with Myotonic Dystrophy and dysphagia (difficulty swallowing).

RESEARCH PROCEDURES: 10-20 caregivers will be recruited by asking a sample of individuals with DM1 who attend the LHSC Neuromuscular Clinic to identify someone they consider a "caregiver". For the purpose of this study, a "caregiver" is defined as a spouse, family member, or friend who participates in helping the individual with DM1 manage his or her symptoms. You are being invited to participate in this study because an individual with DM1 identified you as a caregiver.

If you agree to participate, you will be asked to fill out a short demographic questionnaire and to take part in an interview. During the interview, you will be asked questions about your role as a caregiver, your thoughts about swallowing difficulty experienced by the individual with DM1, your comfort level in responding to a choking episode, and your ability to help you're the individual with DM1 follow safe swallowing recommendations given to him or her by a health care professional. The interview will take approximately 60-90 minutes, but the session may be longer if you have a lot of ideas to share. You may take a break or stop the session if you feel tired.

Once all of the interviews have been completed and analyzed, the study investigators will compile a list of themes that were brought up during the interviews. You will have an opportunity to see this list and to give your opinion about the importance of each theme. This information will ultimately be used to develop programs and resources to help individuals with DM1 and their caregivers monitor and manage safe swallowing.

June 10, 2011

Page 1 of 3

Participant's Initials: _____

CONFIDENTIALITY: Your answers will be kept confidential. Only the study investigators will have access to the answers. All forms and audio tapes will be stored in a locked office, and the interview transcripts will be stored on a password protected computer. At no time will your name be attached to reports or study publications.

BENEFITS: There is no direct benefit to you for participating in this study. Results of this study may contribute to general knowledge about education around, and management of, swallowing difficulty for individuals with Myotonic Dystrophy.

RISKS: There is minimal risk to participating in this study. However, you may feel that some questions are too personal and/or they make you feel sad. You have the right to refuse to answer any question for any reason. If necessary, we can stop the interview. Dr. Venance will be available to talk with you if you have specific concerns about issues that came up during the interview.

VOLUNTARY PARTICIPATION: Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions, or withdraw at any time.

QUESTIONS: To learn more about this study, please contact Kori LaDonna a [REDACTED] [REDACTED] If you have questions about your rights as a research participant, you may contact [REDACTED] Vice President of Research, Lawson Health Research Institute at [REDACTED]

You will be provided with a copy of this Letter of Information and signed consent form for your records.

June 10, 2011

Page 2 of 3

Participant's Initials: _____

**PROTOCOL TITLE: A Pilot Study Assessing the Needs of Caregivers for
Individuals with Myotonic Dystrophy (DM1) and Dysphagia**

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have
read the Letter of Information, have had the nature of the study explained to me, and I
agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

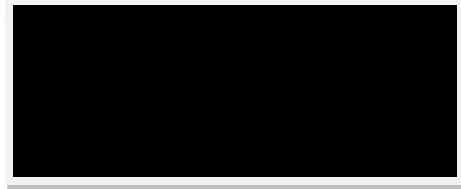
Signature: _____

June 10, 2011

Page 3 of 3

Participant's Initials: _____

Appendix O: Photovoice Study Letter of Information



LETTER OF INFORMATION

Study Title: Picturing the Experience of Living with Myotonic Dystrophy (DM1)

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
 Health and Rehabilitation Sciences (Measurement and Methods)
 University of Western Ontario
 Email: [REDACTED]

Shannon L. Venance, MD, PhD
 Clinical Neurological Sciences
 LHSC-UH
 Phone: [REDACTED]
 Email: [REDACTED]

This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you have been diagnosed with Myotonic Dystrophy (DM1). The purpose of this study is to understand what it is like to live with DM1, and to identify things that help you manage your condition. The researchers are also evaluating a research tool called Photovoice.

Study Information:

Visit 1: If you choose to participate, you will come to the 7th Floor of University Hospital where one of the researchers will describe the study in detail to you. If you give your consent, you will be given a digital camera and instructed on its use. You will be asked to “take pictures of what it is like to live with Myotonic Dystrophy.” You will also be asked to take pictures of things that help you manage your symptoms, or things that make managing your symptoms difficult. You may take as many pictures as you wish. You will also be given a log book in which you can keep track of what you take a picture of, title your photographs, and take notes about your participation in the study. You do not have to write in the log book if you

Participant's Initials: _____

Page 1 of 4

December 14, 2012

choose not to. You will discuss with the researcher things that you might take pictures of, and how to obtain consent from people that you would like to photograph. Finally, you will be asked to complete a short demographic questionnaire. This visit will take approximately one hour, and other people living with DM1 may participate. This visit will be audio tape recorded, but you may request that the recording be stopped at any time. You will be given two to three weeks to take your pictures.

Visit II: The researchers will contact you to schedule an individual interview approximately one month after your first visit. This visit will take place on the 7th floor of University Hospital. Your camera and log book will be collected at this time, and you will have the opportunity to discuss each of your photographs with the researcher. The researcher will also ask you questions about your experience participating in the study. You will be asked to select at least three photographs that you would like to discuss at a focus group. This visit will be audio tape recorded, but you may request that the recording be stopped at any time. This visit will last approximately 1-2 hours.

Visit III: Once you have selected at least three of your photographs, you will have the opportunity to share them with other people participating in the study. The photographs will be projected onto a screen, and you and the other research participants may talk about what you took pictures of, the meaning of your pictures, what you chose not to photograph, and anything else you think is relevant to living with DM1. The researcher will also ask you questions about your experiences participating in the research project. The focus group may have 5-8 people in it, and it may last approximately 2 hours. This visit will be tape recorded, but you may request that the recording be stopped at any time. The researcher will give you information about the location of the focus group when it is scheduled.

Visit IV (Optional): Once you have finished Visits I-III, you have completed the study. However, you may keep your camera and log book for a few extra weeks to take pictures, then return for a follow-up interview and/or focus group. If you choose to take part in this optional portion of the study, the same things that occurred in Visits II or III (depending on whether you choose to attend an individual interview, a focus group, or both) will take place. It is important to remember that you do not have to do this part of the study.

Your photographs are yours to keep. They will be printed and given to you at the end of the study.

Chart Review: The researchers are seeking your permission to review your medical chart to learn more information about your health, your symptoms, and things that you and/or your health care provider(s) consider important. Your health information will remain confidential. You may participate in the study without giving the researchers permission to review your chart.

Participant's Initials: _____

Page 2 of 4

December 14, 2012

Risks:

There are no known risks to participating in this study. However, you may feel uncomfortable talking about certain things related to living with DM1.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals. You are an expert about what it is like to live with DM1, and your experience may enhance the knowledge of the medical and academic community. You will be reimbursed for parking, and light refreshments will be served at each visit.

Confidentiality:

The interviews will be recorded, but you may ask the researcher to stop the recording at any time. You may still participate if you do not want to be recorded during your individual interview. However, the focus groups will be taped. Focus group members are asked to keep everything they hear confidential and not to discuss it outside of the meeting. However, we cannot guarantee that confidentiality will be maintained by group members. You may choose not to attend the focus group. Your name will never be published, and numbers will be used to identify pictures, log books, and transcripts of the recordings. The consent form, pictures, log books, and transcripts of the recordings will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your names will not be used and no information that discloses your identity will be released or published without your permission. On the last page of the consent form, you will be given the opportunity to choose whether or not the researchers can use your photographs for teaching, research, and publication purposes. You can still be in the study if you check "No" to any of these.

Voluntary Participation:

Whether or not you decide to participate in this study is completely voluntary. Deciding not to participate, or choosing to withdraw your consent, will not impact your current or future medical care.

Questions:

Participant's Initials: _____

Page 3 of 4

December 14, 2012



PROTOCOL TITLE: Picturing the Experience of Living with Myotonic Dystrophy (DM1)

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

I give permission to use my photographs for:	YES	NO
Research purposes		
Teaching purposes		
Posters/Publications/Presentations		

Participant's Initials: _____

Page 4 of 4

December 14, 2012


Appendix B: Letter of Information - Individual with DM1





LETTER OF INFORMATION—Individual with DM1

Study Title: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
Health and Rehabilitation Sciences (Measurement and Methods)
University of Western Ontario
Email: 

Shannon L. Venance, MD, PhD
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital
Phone: 
Email: 

Christine Piechowicz, BA
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital
Phone: 
Email: 

This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you have been diagnosed with Myotonic Dystrophy (DM1). The purpose of this study is to understand your reasons for choosing whether or not to receive regular follow-up care at the outpatient neurology clinic.

Page 1 of 4

Participant's Initials _____

January 16, 2012

Study Information:

If you choose to participate, you will come to the 7th floor of University Hospital where you will participate in an interview lasting approximately 1 hour. During the interview, you will be asked questions about your condition, your opinions about the outpatient neurology clinic, and about your clinic attendance. You may have a spouse, family member, or friend present during the interview.

Following your initial interview, you may be asked to return for another interview. This may occur if the researchers have learned new information that they would like to discuss with you, and/or they would like to talk to you about some of the preliminary findings from the research study.

In addition, the researchers would like to review your medical chart to find out information about you, the history of your condition (including genetic testing results, if applicable), medications you take and other things you may do to manage your condition, problems that you bring up with your health care provider, and information about the number of appointments you have attended. You may participate in the interview(s) even if you do not want the researchers to look at your chart.

Risks:

There are no known risks to participating in this study. However, you may feel uncomfortable talking about certain things related to living with your condition.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals. You are an expert about what it is like to live with your condition, and your experience may enhance the knowledge of the medical and academic community. You will be reimbursed for parking.

Confidentiality:

The interview will be recorded, but you may ask the researcher to stop the recording at any time. Numbers will be used to identify transcripts of the recordings, and all transcripts will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. Your name will not be attached to information gathered from your medical chart. While the researchers will make every effort to protect your confidentiality, this cannot be guaranteed. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your names will not be used and no information that discloses your

Page 2 of 4

Participant's Initials _____

January 16, 2012

identity will be released or published without your permission. Your health care provider will not know what you say in your interview.

Voluntary Participation:

Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions or withdraw from the study at any time with no effect on your future care.

Questions:



PROTOCOL TITLE: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

I agree to let the researchers review my medical chart	YES	NO
--	-----	----

Appendix Q: Letter of Information - Individual with Huntington's Disease



LETTER OF INFORMATION—Individuals with HD

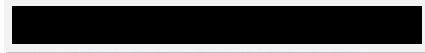
Study Title: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
Health and Rehabilitation Sciences (Measurement and Methods)
University of Western Ontario



Shannon L. Venance, MD, PhD
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



Christine Piechowicz, BA
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you have been diagnosed with Huntington's disease (HD). The purpose of this study is to understand your reasons for choosing whether or not to receive regular follow-up care at the outpatient neurology clinic.

Page 1 of 4

Participant's Initials _____

June 13, 2012

Study Information:

lasting approximately 1 hour. During the interview, you will be asked questions about your condition, your opinions about the outpatient neurology clinic, and about your clinic attendance. You may have a spouse, family member, or friend present during the interview.

Following your initial interview, you may be asked to participate in another interview. This may occur if the researchers have learned new information that they would like to discuss with you, and/or they would like to talk to you about some of the preliminary findings from the research study.

In addition, the researchers would like to review your medical chart to find out information about you, the history of your condition (including genetic testing results, if applicable), medications you take and other things you may do to manage your condition, problems that you bring up with your health care provider, and information about the number of appointments you have attended. You may participate in the interview(s) even if you do not want the researchers to look at your chart.

Risks:

There are no known risks to participating in this study. However, you may feel uncomfortable talking about certain things related to living with your condition.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals. You are an expert about what it is like to live with your condition, and your experience may enhance the knowledge of the medical and academic community. You will be reimbursed for parking.

Confidentiality:

The interview will be recorded, but you may ask the researcher to stop the recording at any time. Numbers will be used to identify transcripts of the recordings, and all transcripts will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. Your name will not be attached to information gathered from your medical chart. While the researchers will make every effort to protect your confidentiality, this cannot be guaranteed. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your names will not be used and no information that discloses your

Page 2 of 4

Participant's Initials _____

June 13, 2012

identity will be released or published without your permission. Your health care provider will not know what you say in your interview.

Voluntary Participation:

Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions or withdraw from the study at any time with no effect on your future care.

Questions:



PROTOCOL TITLE: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

I agree to let the researchers review my medical chart	YES	NO
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Appendix B: Letter of Information – Caregiver of Individual with DM1

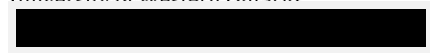


LETTER OF INFORMATION—Caregiver for Individual with DM1

Study Title: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
Health and Rehabilitation Sciences (Measurement and Methods)
University of Western Ontario



Shannon L. Venance, MD, PhD
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



Christine Piechowicz, BA
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you are a family member, friend, or loved one of someone with Myotonic Dystrophy (DM1). The purpose of this study is to understand the reasons the individual with DM1 chooses whether or not to receive regular follow-up care at the outpatient neurology clinic and his/her expectations for care. You are being asked to provide supplemental opinions or information regarding clinical care.

Page 1 of 4

Participant's Initials _____

April 10, 2012

Study Information:

If you choose to participate, you will come to the 7th floor of University Hospital where you will participate in an interview lasting approximately 1 hour. During the interview, you will be asked questions about your loved one's condition, your opinions about the outpatient neurology clinic, and about your loved one's clinic attendance.

Following your initial interview, you may be asked to participate in another interview. This may occur if the researchers have learned new information that they would like to discuss with you, and/or they would like to talk to you about some of the preliminary findings from the research study.

Risks:

There are no known risks to participating in this study. However, you may feel uncomfortable talking about certain things related to Myotonic dystrophy.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals. The information you provide may enhance the knowledge of the medical and academic community. You will be reimbursed for parking.

Confidentiality:

The interview will be recorded, but you may ask the researcher to stop the recording at any time. Numbers will be used to identify transcripts of the recordings, and all transcripts will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. While the researchers will make every effort to protect your confidentiality, this cannot be guaranteed. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your name will not be used and no information that discloses your identity will be released or published without your permission. Your loved one's health care provider will not know what you say in your interview.

Voluntary Participation:

Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions or withdraw from the study at any time with no effect on your future care.

Questions:

PROTOCOL TITLE: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

Page 4 of 4

Participant's Initials _____

April 10, 2012

Appendix S: Letter of Information - Caregiver of Individual with Huntington's Disease



LETTER OF INFORMATION—Caregiver for Individual with HD

Study Title: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
Health and Rehabilitation Sciences (Measurement and Methods)
University of Western Ontario



Shannon L. Venance, MD, PhD
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



Christine Piechowicz, BA
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you are a family member, friend, or loved one of someone with Huntington's disease (HD). The purpose of this study is to understand the reasons the individual with HD chooses whether or not to receive regular follow-up care at the outpatient neurology clinic and his/her expectations for care. You are being asked to provide supplemental opinions or information regarding clinical care.

Page 1 of 4

Participant's Initials _____

April 10, 2012

Study Information:

If you choose to participate, you will come to the 7th floor of University Hospital where you will participate in an interview lasting approximately 1 hour. During the interview, you will be asked questions about your loved one's condition, your opinions about the outpatient neurology clinic, and about your loved one's clinic attendance.

Following your initial interview, you may be asked to participate in another interview. This may occur if the researchers have learned new information that they would like to discuss with you, and/or they would like to talk to you about some of the preliminary findings from the research study.

Risks:

There are no known risks to participating in this study. However, you may feel uncomfortable talking about certain things related to Huntington's disease.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals. The information you provide may enhance the knowledge of the medical and academic community. You will be reimbursed for parking.

Confidentiality:

The interview will be recorded, but you may ask the researcher to stop the recording at any time. Numbers will be used to identify transcripts of the recordings, and all transcripts will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. While the researchers will make every effort to protect your confidentiality, this cannot be guaranteed. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your name will not be used and no information that discloses your identity will be released or published without your permission. Your loved one's health care provider will not know what you say in your interview.

Voluntary Participation:

Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions or withdraw from the study at any time with no effect on your future care.

Questions:

Participant's Initials _____

Page 3 of 4

April 10, 2012

PROTOCOL TITLE: Understanding the Decision-Making Process for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

Page 4 of 4

Participant's Initials _____

April 10, 2012

Appendix T: Letter of Information - Health Care Practitioner for Individual with DM1



LETTER OF INFORMATION—Health Care Provider (DM1)

Study Title: Understanding the Decision-Making Processes for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
Health and Rehabilitation Sciences (Measurement and Methods)
University of Western Ontario



Shannon L. Venance, MD, PhD
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



Christine Piechowicz, BA
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you are a health care provider for individuals with Myotonic Dystrophy (DM1). The purpose of this study is to understand your beliefs about the reasons these individuals do or do not choose to receive on-going care at

Page 1 of 4

Participant's Initials: _____

January 16, 2012

the outpatient neurology clinic. We would also like to understand your goals for the care of these individuals.

Study Information:

If you choose to participate, you will participate in an interview lasting approximately 1 hour. The interview will take place in a location that is convenient for you. During the interview, you will be asked questions about DM1, your opinions about clinic attendance for these individuals, and about your goals for their care.

Following your initial interview, you may be asked to participate in another interview. This may occur if the researchers have learned new information that they would like to discuss with you, and/or they would like to talk to you about some of the preliminary findings from the research study.

With your permission, and with the consent of your patients, the researchers would like to access the charts of individuals participating in this research study. In particular, the researchers are interested in information regarding patient demographics, disease history and management, issues of concern to both you and your patients, and appointment-keeping behavior. You may participate in the study even if you do not want the researchers to have access to the charts.

Risks:

There are no known risks to participating in this study.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals, and the results may enhance the knowledge of the medical and academic communities.

Confidentiality:

The interview will be recorded, but you may ask the researcher to stop the recording at any time. Numbers will be used to identify transcripts of the recordings, and all transcripts will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your

names will not be used and no information that discloses your identity will be released or published without your permission.

Voluntary Participation:

Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions or withdraw from the study at any time with no effect on your future care.

Questions:



PROTOCOL TITLE: Exploring the Motivations and Decision-Making Processes for Individuals with Chronic Neurological Conditions who Choose to Receive On-Going Care at an Outpatient Neurology Clinic

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

If my patients consent, I agree to give the researchers access to the patients' medical charts.	YES	NO
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Appendix U: Letter of Information - Health Care Practitioner for Individual with Huntington's Disease



LETTER OF INFORMATION—Health Care Provider (HD)

Study Title: Understanding the Decision-Making Processes for Individuals with Chronic Neurological Conditions Regarding On-Going Care at an Outpatient Neurology Clinic

Study Investigators:

Kori A. LaDonna, BA, PhD Candidate
Health and Rehabilitation Sciences (Measurement and Methods)
University of Western Ontario



Shannon L. Venance, MD, PhD
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



Christine Piechowicz, BA
Clinical Neurological Sciences
London Health Sciences Centre—University Hospital



This research project is being conducted by researchers at the University of Western Ontario and at London Health Sciences Centre. You have been invited to participate in this research study because you are a health care provider for individuals with Huntington's disease (HD). The purpose of this study is to understand your beliefs about the reasons these individuals do or do not choose to receive on-going care at

Page 1 of 4

Participant's Initials: _____

January 16, 2012

the outpatient neurology clinic. We would also like to understand your goals for the care of these individuals.

Study Information:

If you choose to participate, you will participate in an interview lasting approximately 1 hour. The interview will take place in a location that is convenient for you. During the interview, you will be asked questions about HD, your opinions about clinic attendance for these individuals, and about your goals for their care.

Following your initial interview, you may be asked to participate in another interview. This may occur if the researchers have learned new information that they would like to discuss with you, and/or they would like to talk to you about some of the preliminary findings from the research study.

With your permission, and with the consent of your patients, the researchers would like to access the charts of individuals participating in this research study. In particular, the researchers are interested in information regarding patient demographics, disease history and management, issues of concern to both you and your patients, and appointment-keeping behavior. You may participate in the study even if you do not want the researchers to have access to the charts.

Risks:

There are no known risks to participating in this study.

Benefits:

The results of this research will be presented at meetings, presentations, and in scholarly journals, and the results may enhance the knowledge of the medical and academic communities.

Confidentiality:

The interview will be recorded, but you may ask the researcher to stop the recording at any time. Numbers will be used to identify transcripts of the recordings, and all transcripts will be locked in a secure place at the University of Western Ontario and kept for future consultation by the researchers. Representatives of the University of Western Ontario Health Sciences Research Ethics Board may contact you, or require access to your study-related records to monitor the conduct of the research. If the results of the study are published, your names will not be used and no information that discloses your identity will be released or published without your permission.

Page 2 of 4

Participant's Initials: _____

January 16, 2012

Voluntary Participation:

Participation in this study is voluntary. You may refuse to participate, refuse to answer any questions or withdraw from the study at any time with no effect on your future care.

Questions:

PROTOCOL TITLE: Exploring the Motivations and Decision-Making Processes for Individuals with Chronic Neurological Conditions who Choose to Receive On-Going Care at an Outpatient Neurology Clinic

CONSENT FOR PARTICIPATION IN RESEARCH

I _____ (YOUR NAME) have read the Letter of Information, have had the nature of the study explained to me, and I agree to participate. All questions have been answered to my satisfaction.

Name of Participant: _____ (PRINT NAME)

Signature of Participant: _____ (SIGNATURE)

Date: _____

PERSON OBTAINING CONSENT

Print Name and Title: _____

Signature: _____

If my patients consent, I agree to give the researchers access to the patients' medical charts.	YES	NO
---	-----	----

Curriculum Vitae

Name:	Kori A. LaDonna
Post-secondary Education and Degrees:	<p>State University of New York at Geneseo Geneseo, New York, USA 1999-2001 BA, Communication and BA, English</p> <p>University of Western Ontario London, Ontario, Canada 2005 Certificate in Clinical Trials Management</p> <p>University of Western Ontario London, Ontario, Canada 2006-2007 Health and Social Sciences</p> <p>University of Western Ontario London, Ontario, Canada 2007-2008 Masters of Science in Health Promotion Candidate</p> <p>University of Western Ontario London, Ontario, Canada 2008-Present Health and Rehabilitation Sciences PhD Candidate</p>
Related Work Experience	<p>Support Group Facilitator Huntington's Disease Support Group, Rochester, NY 2001-2005</p> <p>Intern, Huntington's Disease Society of America's Center of Excellence at the University of Rochester, Rochester, NY</p>

2001

Research Assistant/Clinical Coordinator

Cognitive and Behavioral Neurology Unit, University of
Rochester, Rochester, NY

2002-2004

Support Group Facilitator

Young Parkinson's Disease Support Group, Rochester, NY

2003-2004

Research Assistant/Clinical Coordinator

Department of Clinical Neurological Sciences, London Health
Sciences Centre, London, Ontario

2006-2008

Research Associate

Centre for Education Research & Innovation, Western University
London, Ontario

2013-Present

Publications:

Richard IH, Frank SA, McDermott MP, Wang H, Justus AW, **LaDonna KA**, Kurlan R. (2004) The Ups and Downs of Parkinson's Disease: A prospective study of mood and anxiety fluctuations. *Cognitive and Behavioral Neurology*. 2004 (17): 201-207.

Richard IH, Frank SA, **LaDonna KA**, Wang H, McDermott MP, Kurlan R. (2005) Mood Fluctuations in Parkinson's Disease: A Pilot Study Comparing the Effects of Intravenous and Oral Levodopa Administration. *Neuropsychiatric Disease and Treatment*. 1(3): 261-268.

Richard IH, **LaDonna KA**, Hartman R, Podgorski C, Kurlan R, SAD-PD Study Group. (2007) The patient's perspective: Results of a survey assessing knowledge about and attitudes toward depression in PD. *Neuropsychiatric Disease and Treatment*. 3(6): 903-906.

LaDonna KA. (2011) A literature review of studies using qualitative research to explore Chronic Neuromuscular Disease. *Journal of Neuroscience Nurses*, 43(3): 172-182.

LaDonna KA, Koopman WJ & Venance SL. (2011) Myotonic dystrophy (DM1) and dysphagia: the need for dysphagia management guidelines and an assessment tool. *Canadian Journal of Neuroscience Nursing*, 33(1): 42-46.

LaDonna KA & Ravenek, MJ. (2014) Challenges & strategies for conducting qualitative research with individuals with rare movement disorders. *The Qualitative Report*(19), 31: 1-18.

Venance SL, **LaDonna KA** & Watling CJ. (2014) Exploring front-line faculty perspectives on curriculum change. *Medical Education*.

LaDonna KA, Koopman WJ, Ray SL & Venance SL. (In press) Hard to swallow: A phenomenological exploration of the experience of caring for individuals with Myotonic Dystrophy (DM1) and Dysphagia. *Journal of Neuroscience Nursing*.

LaDonna KA & Venance SL. (In press) Picturing the experience of living with Myotonic Dystrophy (DM1): A qualitative exploration using Photovoice. *Journal of Neuroscience Nursing*.

LaDonna, KA, Ray, SL, Watling, CJ, Piechowicz C, & Venance SL. (In preparation) Perceptions of health care professionals caring for patients with chronic, progressive hereditary and life-limiting neurological conditions: "We Like to Think We're Making a Difference"

LaDonna, KA, Ray, SL, Watling, CJ, Piechowicz C, & Venance SL. (In preparation) '[The neurologist] is throwing you a raft': Exploring motivations for on-going clinic

attendance for individuals living with chronic, progressive and life-limiting neurological conditions.