Myotonic dystrophy Type 1: exploring the health care experiences of people living with DM1. A Research Protocol.

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This article outlines a research protocol for investigating the health care experiences of people living with Myotonic Dystrophy Type 1 (Dytrophia Myotonica; DM1).
Introduction: DM1 is a neuromuscular condition of highly variable presentation across the lifespan. It is a complex and variable condition which impacts many body systems. Care consensus, clinical experience and scientific genetic findings dominate much of the literature. Findings from the literature indicate health care input from multiple disciplines is recommended to help identify and prevent or minimise disability and early mortality. It is these recommendations for intense health care input and the perceptions people living with DM1 have of their health care that have prompted this enquiry.

Objectives: Investigate the experiences of health care from 3 different groups: people living with DM1, their family members, carers or support people, and health care providers. Potentially illuminating what we as nurses and health care providers do well or not so well.

Intended methods: Guided by Merleau-Ponty's phenomenology of perception and triangulate these gathered experiences to ascertain if there is convergence or divergence between health care experiences of recipients and providers. This research will utilise a qualitative approach with methodology drawn from phenomenology. Semi-structured Interviews will be conducted with 3 different groups to gain insight into perceptions, experiences, and outcomes of health care interactions. People living with DM1, family members, carers and friends and health professionals providing services and care for people living with DM1 will be invited to participate.

Keywords: Myotonic Dystrophy Type 1, Health care provision, patient experience

Background

Myotonic Dystrophy Type 1- the condition

Prevalence studies conducted have concluded that approximately 1:8000 people live with DM1 (Theadom et al., 2014; Gagnon et al., 2010; De Antonio et al., 2016). The condition can affect people across the lifespan and highly variable symptom presentations are evident. Several classifications exist including a congenital form, childhood onset and classic adult onset. The earlier the onset or diagnosis of DM1, the more debilitating the symptoms.

Impact of living with DM1

People with DM1 are at risk of sudden death. The median age of death is 60 years for males and 59 years for females. Death is usually due to cardiac arrythmias, respiratory failure and cancers. The condition presents with highly variable complex symptoms.

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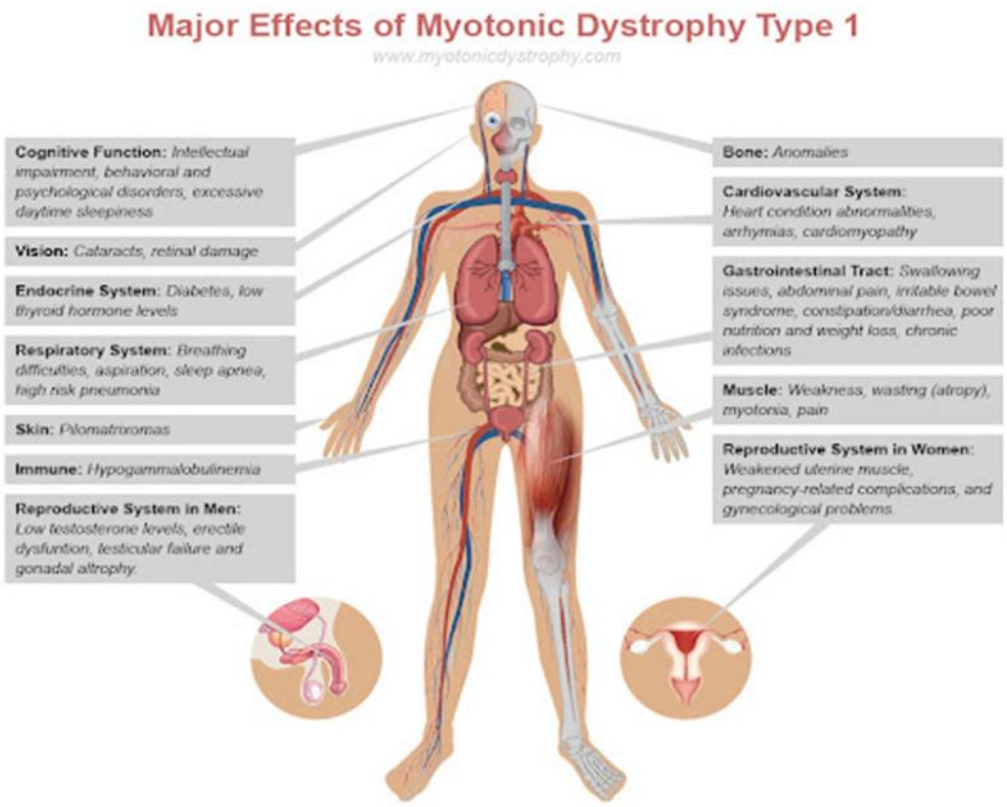

Figure 1: Major effects of DM1 http://myotonicdystrophy.com/http:/mytonicdystrophy.com/general-information/ retrieved 06.11.2021

However there will be people with genetically identified DM1 who remain asymptomatic and others who will be severely affected (de Die-Smulders et al, 1998; Turner, Hilton-Jones, 2008). The literature also describes a temperament and personality character specific to DM1. Again these characteristics do not apply to all people living with DM1 and debate continues as to how influential socio-economic and educational status are on character and temperament development (Winbald et al, 2005; Bertrand et al, 2015; Baldanzi et al, 2016).

Genetics of DM1

For more than a century, the inheritance of DM1 has been recognized. Hans Steinert described the condition back in 1909. It was not until 1992 that formal identification was reported involving the cytosine-thymine-guanine (CTG) trinucleotide repeats on the dystrophia myotonica protein kinase (DMPK) gene (Groh et al., 2011; Jansen et al., 1992). The scientific literature reports a positive association between high CTG repeats and the severity of clinical symptoms.

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Figure 1 shows the multiple body systems impacted by DM1 and consequently the detrimental effects these can have on quality of life (Turner & Hilton-Jones, 2010; Winblad et al, 2005; Laberge, 2007). Currently there is no cure for DM1, there are no available neuroprotective treatments, surveillance of cardiac and respiratory function is lifelong and frequent (6 monthly if there are pathological findings requiring monitoring). There is a growing body of evidence that allied health professional input from physiotherapists, speech pathologists and occupational therapists can have a positive impact on symptom management (Raymond et al, 2016; Boentert et al., 2020).

Literature Review

People living with DM1 have complex health care needs often requiring input from multiple specialties for multiple body systems (Ashizawa et al., 2018; Chouinard, 2008; White, 2020). There is a large amount of scientifically based research focusing on body system effects, genetic identification, and impact. There are survey results and self-reported rating scales describing activities of daily living and quality of life for people with DM1 (Landfeldt et al., 2020; Nätterlund & Ahlström, 1999; Raymond et al, 2019; Peric et al., 2016). Several qualitative research projects explore living with DM1 and the effect on everyday activities, personal perspectives from individuals as well as partners and health care providers experiences of providing care for people with DM1 (LaDonna et al, 2016, 2017; Cup et al., 2011; Magliano & Politano, 2016).

There are published consensus-based recommendations for DM1 care from clinical experts with experience in DM1 care. These opinion articles clearly state that due to a current lack of studies and research required to produce evidence-based guidelines consensus-based guidelines have been developed to help standardise care (Ashizawa et al., 2018; Chouinard, 2008; White, 2020; Gagnon et al., 2010). Clearly lacking in the current body of DM1 literature is an Australian perspective of people living with DM1 as recipients of health care. There is report and a call for care standards for children living with congenital DM and youths living with neuromuscular conditions as a single cohort rather than specific conditions such as DM1 (Travlos et al, 2016; Yang et al, 2018). A survey was conducted of neuromuscular clinic attendees with the demographic data showing several people with DM1 took part, but their specific health care needs weren't addressed (Anderson et al., 2018).

There is a need for education for patients and families throughout the disease course, education for health care providers, translating recommended care into current clinical care (Raymond et al, 2016; Hartley et al, 2011). Other research has recommended further work be done that focusses on DM1 symptoms that have clinical and personal impact so these can be recognised by both health professionals and people with DM1. Discovering this could lead to creating a shared decision making and person-centred care approach in the clinical environment (Lecordier et al, 2017; LaDonna et al, 2017, 2016).

Research Proposal

The following research protocol has been developed to guide a deeper enquiry of how health care is experienced by people living with DM1.

Research Aims

To listen to the health care experiences of Australian people living with DM1. To listen to the relatives / carers / friends' experiences of keeping their family member / care recipient engaged in recommended health care activities. To gain the perspectives and experiences of health care professionals providing care to people living with DM1.

Research Rationale

DM1 remains under identified and poorly understood within the greater health care community despite people living with DM1 often presenting with complex and variable health and social care needs that require input from multiple disciplines. Importantly, health professionals and people living with DM1 at times describe engaging in health care a challenge.

Objectives

Understand the health care experiences of people living with DM1.

Research Question

What are the health care experiences of Australian people living with DM1?

Recruitment

The research will be advertised with the assistance of Muscular Dystrophy Queensland (MDQ) the peak body in Queensland providing support through services and information to members and their families living with myotonic dystrophy as well as other neuromuscular conditions.

The investigators intend to approach other Australian Neuromuscular clinics to invite health professionals and patients to participate.

Snowballing strategies will be used to encourage participation from those who may not have access to advertised information.

Participants

Participants will be invited to take part and comprise three different groups.

Table 1

Group 1 People living with DM1 diagnosed by a medical specialist by positive genetic testing and/or recognised family inheritance.	
Inclusion criteria	**Exclusion criteria**
Aged 18 years or over. Be able to speak, read and write in English. Able to give consent to participate on their own behalf. Have attended a health care appointment in the previous 12 months. Be willing and able to take part in a recorded interview for approximately 1 hour.	Under the age of 18 years. Have a known cognitive impairment that impacts on their ability to provide informed consent on their own behalf.

Table 2

Group 2 Support people: family, carers, friends of a person living with DM1	
Inclusion Criteria	**Exclusion criteria**
Aged 18 years or over. Be able to speak, read and write in English. Able to give consent to participate on their own behalf. Have attended a health care appointment supporting a person with DM1 in the previous 12 months.	Under the age of 18 years. Has not assisted a person with DM1 in health care participation.

Table 3

Group 3 Health care professionals	
Inclusion Criteria	**Exclusion criteria**
Have provided care to people living with DM1 in the past 12 months. Provided care directly related to the DM1 condition – e.g., symptom management, symptom monitoring (cardiac or respiratory tests, physical therapy). Have greater than 12 months experience in health care delivery for people living with DM1.	Less than 12 months experience of DM1 care provision.

protection will be ensured by using research data management storage. This secure platform

Participant numbers

It is intended that a minimum of 5 participants

from each group will take part in semi-structured interviews. However, recruitment will continue with more participants invited to take part in interviews until no new content emerges or until data saturation is reached (Creswell, 2013, p. 155; Malterud, Siersma, & Guassora, 2015; Sandelowski, 1995).

Methodology

The most appropriate method to answer the research question is to use a qualitative approach. The philosophical framework of this research will follow an Interpretive paradigm. A Phenomenological approach will be taken (Merleau-Ponty 1945, 2013; Dowling, 2007). The theoretical framework to guide the analysis of the interviews will be Albert Bandura's Theory of Self Efficacy / Self-Management. Self-efficacy is gained through mastery over stressful situations and become a generalisable skill that can be used in future situations. This mastery can be demonstrated in how people respond to performance accomplishments. Accomplishments are achieved by responses to vicarious experience, verbal persuasion, and emotional arousal. Focusing on how people living with DM1 demonstrate mastery over their health care experiences may highlight their self-efficacy skills (Bandura, 1978).

Methods

Guided by Merleau-Ponty's phenomenology of perception triangulation of the gathered experiences from the 3 groups will occur to uncover any convergence or divergence between them. Interested potential participants will be provided an explanation of the research and provided information and consent forms. Prior to enrolment the DM1 group will be administered the University of California, San Diego Brief Assessment of Capacity to Consent (UBACC©) to ensure they have full understanding of the research project and what their role as participant is so they can provide informed written consent on their own behalf. Semi structured interviews will be conducted following a predetermined interview guide relevant to the group the participant belongs in. All interviews will be audio recorded. Intended interview length is 1 hour.

Data Analysis

Thematic analysis will be performed following Braun & Clarke's guidelines (Braun & Clarke, 2006)

1. Data familiarisation: reading and re-reading interview transcripts.
2. Generate codes: phrases or keywords that summarise a concept or experience, apply codes throughout a transcript.
3. Allocate themes: apply a broader concept

that codes can fit into.

4. Share codes & themes with co-investigators and experienced peers.

5. Definitions and names will be allocated to each identified theme.

6. Ensure identified codes fit an allocated themes then generated a report using clear definitions of each theme.

Ethical Considerations

Ethical (HREC) approval has been granted by the researchers' tertiary institutional committee. Recruitment information will inform potential participants they can take part in interviews at their convenience. These can be conducted either by phone, teleconferencing or face to face. It is important that the co-investigator conducting the interviews ensures that clinical and research roles remain separate. This addresses any potential power bias that may occur if participants have a clinician / patient relationship. Signed informed consent is required from all participants prior to taking part in the research. Complaint management has been addressed with contact number and email address provided for the approving HREC coordinator. Contact details of appropriate counselling services have been provided in the participant information should participants experience any distress when discussing their healthcare experiences during an interview.

All participants will be de-identified by allocating pseudonyms and the names of health care facilities identified during interviews will be removed from transcripts. This is to ensure and maintain confidentiality of all participants. Data management and is maintained by the University of Queensland and will be used for storage of all participant demographics, interview transcripts and data analysis.

Practical considerations

Options of phone, on-line teleconferencing (Zoom™, Facetime™ or Microsoft teams™ platforms per participants preference), and face to face interviews will be offered. Interviews will be audio recorded and manually transcribed or uploaded to transcription software as interview numbers increase.

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