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Capturing Health-Related Quality of Life in Adults with Charcot-Marie-Tooth Disease: A Digital Photovoice Investigation

Elaine Walklet

A thesis submitted in partial fulfilment of the requirements of the University of the West of England, Bristol for the degree of Professional Doctorate in Health Psychology

Faculty of Health and Applied Sciences, University of the West of England, Bristol
February 2018

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Introduction to the thesis

The following thesis has been produced to fulfil the research component of the Professional Doctorate in Health Psychology. It therefore comprises of a systematic review and an empirical study; both of which focus on adults with neuromuscular disease and quality of life.

The systematic review is entitled: “Psychosocial interventions for quality of life and well-being in adults with neuromuscular conditions: a systematic review and narrative synthesis”. This systematic review was completed in year 1 of a taught doctoral course and published in year 2. The original submission and a link to the published article can both be found in Appendix A. One of the findings of the systematic review was that measurement of quality of life was inconsistent across studies. Furthermore, within individual intervention studies, different measures of quality of life produced different outcomes. This complicated interpretation of the findings and raised a question about whether quality of life was being validly assessed. Thus, the starting point for the empirical study was the need to better understand quality of life within this population group, with a view to informing future development of condition specific measures and targeted interventions. A detailed contents page can be found overleaf.
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Acknowledgements

First and foremost, I would like to thank the participants of this study. Without you, this research would not have been possible. Thank you for taking the time to capture your experiences and for reflecting on them with such honesty. It was a privilege to analyse the data you provided and I am so grateful to you all for taking part.

Next, thank you to my fantastic supervisory team. To my Director of Studies Dr Tim Moss – thank you for always believing in me and for supporting me to find my own way and have confidence in myself. Professor Nicki Walsh and Professor Eleanor Bradley – thank you for your insightful guidance and feedback. It has been a pleasure working with you all.

Thank you to my examiners Dr Aimee Aubeeluck and Dr Heidi Williamson for making my viva an enjoyable experience and for the helpful feedback and advice you gave me.

Thank you to my patient partner Karen Butcher for your support at every stage of this research. You are truly an inspiration in everything you do to support people with CMT.

To Sophie Williams – thank you for introducing me to photovoice and inspiring me to use this method. It has been a wonderful journey which may never have happened if I had not attended your conference presentation! Thank you so much for the advice and guidance.
To my fiancée James – thank you for your unwavering belief in me, for the endless cups of tea and for helping me to stay (relatively) calm over the last three years. I could not have done this without you.

To my work colleagues and friends – thank you for encouraging me to complete this course in the first place and for your continued support. In particular, thanks to Dr Kate Muse, Dave Wager, Libby Symonds and Dr Berenice Mahoney.

To my family and friends – thank you for your endless encouragement and for putting up with me over the last three years. In particular thanks to my parents Annette and Malcolm Walklet who have always supported me. To my fellow trainees – thank you for the memories! I feel so lucky to have studied with such an inspiring and supportive group of women. Thank you for everything: Clare Hamlet, Fiona Cunningham, Jo Webb, Laura Mills, Puja Chandegra, Shareen Ali
Definition of Key Terms

Charcot-Marie-Tooth Disease (CMT): CMT is the most common inherited neuromuscular condition in the world. There are various types of CMT which all cause peripheral neuropathy (damage to nerves in the peripheral nervous system) and are associated with a range of degenerative symptoms such as muscle weakness, deformity, instability and chronic pain (Hilton-Jones, Freebody and Stein, 2011).

Health-Related Quality of Life (HRQoL): It is acknowledged that HRQoL is a contested term. For the purposes of this thesis, HRQoL is considered a broad concept which refers to “quality of life relative to one’s health or disease status” (Bakas et al., 2012, p. 1). A subjective, dynamic and multi-faceted construct, HRQoL incorporates physical, psychological, social and spiritual domains (Bakas et al., 2012). Elaboration of this definition can be found on page 17.

Photovoice: Photovoice originates within sociology and is traditionally used as a tool for participatory action research. In the original photovoice method, participants are provided with a disposable camera and asked to photograph aspects of their experience. The photographs form the basis for subsequent critical discussion, which ultimately aims to influence and enhance the community (Wang and Burris, 1997). In this research, the photovoice method incorporates digital photographs and accompanying textual narratives.
List of Abbreviations

AFO = Ankle Foot Orthosis

ASO = Antisense Oligonucleotides

CMT = Charcot-Marie-Tooth Disease

HSMN = Hereditary Sensory and Motor Neuropathy

HRQoL = Health-related Quality of Life

PROMS = Patient-reported outcome measures

QoL = Quality of Life
Abstract

**Rationale:** CMT is a progressive and debilitating neuromuscular condition which has previously been linked with reduced HRQoL. However, little is known about what adults with CMT think about their HRQoL and the various factors that influence it. An in-depth understanding of experience is essential for valid assessment of HRQoL and appropriate intervention.

**Aim:** The aim of this study was to explore how adults living with CMT understand HRQoL and the factors which influence this.

**Method:** A digital photovoice study was conducted. Eleven participants with varying types of CMT completed an online journal over a 2-week period. Participant were asked to photograph anything which they felt influenced their quality of life (positively, negatively, or neutrally) and to write an accompanying reflective narrative. On average, participants posted 8.36 journal entries (including at least one photo and a narrative reflection) during the 2-week study duration.

**Findings:** Thematic analysis of the textual and visual data identified 6 main themes: “The challenge of the every-day”, “Maintaining independence: adaption, adjustment and support”, “Loss of control and future-orientated anxiety”, “CMT Awareness”, “Threats to identity and self-esteem” and “Resilience and positive growth”. Overall, HRQoL emerged as a complex, individual and multi-faceted construct which is influenced by a range of internal and external factors.
**Conclusions:** Whilst the physical aspects of CMT are inherently challenging, threats to HRQoL also arise from the physical and social environment and the psychological impact of living with a progressive, hereditary condition. Despite much adversity, all participants demonstrated substantial resilience and positive growth which helped them to manage the condition and enhanced their HRQoL. A CMT-specific measure of HRQoL and targeted interventions are now urgently required.
1. Introduction

1.1 Charcot-Marie-Tooth Disease

1.1.1 Definition and prevalence. Charcot-Marie-Tooth (CMT) disease is a slowly progressive condition which causes neuropathy of the sensory and motor nerves of the peripheral nervous system (Hilton-Jones, Freebody and Stein, 2011). The name ‘Charcot-Marie-Tooth’ originates from the three doctors who first discovered it: Jean Charcot, Pierre Marie and Howard Henry Tooth. Recently, the condition has also been referred to as Hereditary Motor and Sensory Neuropathy (HSMN), however most patients prefer the name CMT (McCorquodale, Pucillo and Johnson, 2016; Hilton-Jones, Freebody and Stein, 2011).

CMT or HSMN is the most common inherited neuromuscular disorder (Hilton-Jones, Freebody and Stein, 2011) and one of the most commonly inherited disorders in humans (McCorquodale, Pucillo and Johnson, 2016). Approximately 25,000 people live with CMT in the UK and it is estimated that the overall prevalence rate is 1:2500 (CMT UK, 2017; Hilton-Jones, Freebody and Stein, 2011). Despite having a much higher prevalence rate than other neuromuscular disorders such as Duchenne Muscular Dystrophy (Ryder et al., 2017) and Motor Neurone Disease (Mehta et al., 2016), general awareness of CMT is limited (Lafarge et al., 2014).
1.1.2 Clinical presentation. CMT is clinically heterogeneous, with age of onset, disease severity and speed of progression varying significantly between individuals, and within families (Redmond, Burns and Ouvrier, 2008). Typically the condition emerges in adolescence or early adulthood, however, symptoms can present in early childhood or later in life (Yagerman et al., 2012; Hilton-Jones, Freebody & Stein, 2011; Souayah et al., 2007).

CMT is associated with a range of physical symptoms. Typically, it causes reduced sensation, muscle weakness and wasting to distal muscles, usually starting with the feet and legs and, latterly, progressing to the hands (Hilton-Jones, Freebody and Stein, 2011). Muscle weakness and wasting often causes deformities in the feet and hands (e.g. high arches, foot drop, hammer toes, claw-hand etc.) and can lead to an abnormal gait, difficulty with walking and balance, frequent falls, pain and fatigue (McCorquodale, Pucillo and Johnson, 2016; Ramdharry et al., 2012; Boentert et al., 2010; Pareyson and Marchesi, 2009; Padua et al., 2007). Fine motor skills and dexterity can also be significantly impaired once the muscles in the hands are affected (Hilton-Jones, Freebody and Stein, 2011). Research indicates that individuals with CMT often present with additional symptoms to the ‘classic’ CMT profile. These include, but are not limited to, scoliosis of the spine, hip dysplasia, restless leg syndrome, fasciculations (involuntary muscle twitching), cold-induced hand cramp, facial weakness and respiratory insufficiency (Werheid et al., 2016; Yagerman et al., 2012). Thus, the symptom profile of CMT is varied and complex. Accordingly, functional impairment can range from mild to severe and complicated, with some individuals requiring a
wheelchair from an early age (McCorquodale, Pucillo and Johnson, 2016; Padua et al., 2006).

1.1.3 Inheritance pattern and types of CMT. Genetically heterogeneous, CMT includes a group of neuropathies with varying inheritance patterns (autosomal dominant, autosomal recessive or x-linked). Most forms of CMT are autosomal dominant (Vallat and Mathis, 2014), meaning there is a 50% chance of an affected parent passing on the condition to their children. In contrast, recessive forms of CMT (rarer) require both parents to be a carrier for the mutation to be inherited. In this case, each child has a 25% chance of developing the condition and a 50% chance they will be a carrier. X-linked inheritance patterns are more common than recessive and are sex-linked (CMT UK, 2017). In addition to known inheritance patterns, CMT can also develop from a new genetic mutation with no known family history (Vallat and Mathis, 2014).

Over 60 genes have been linked with CMT to date, although it is estimated that there may be up to 100 (Mathis et al., 2015; Taniguchi et al., 2013). There are two predominant types of the condition: type 1 (demyelinating – affects the myelin sheath around the nerve) and type 2 (axonal – directly affects the axon of the nerve), each with numerous subtypes (McCorquodale, Pucillo and Johnson, 2016; Arnold, McEntagart and Younger, 2005). Whilst other types of CMT do exist (e.g. CMT4, CMTX), they are much less common (Mathis et al., 2015). The majority of individuals
diagnosed with CMT have type 1 (50-80%), with a smaller minority having type 2 (10-15%) (McCorquodale, Pucillo and Johnson, 2016; Hilton-Jones, Freebody and Stein, 2011). There are genetic and clinical differences between type 1 and type 2 (McCorquodale, Pucillo and Johnson, 2016). Consequently, CMT is a complex spectrum of disorders.

1.1.4 Diagnosis. Although some people are diagnosed at a relatively young age, many individuals live with the condition for a number of years before a diagnosis is made (Arnold, McEntagart and Younger, 2005). Diagnosis of CMT is achieved via clinical examination of typical signs and symptoms of CMT, electrophysiological tests of nerve function and genetic testing (Pareyson and Marchesi, 2009). Genetic testing is suggested to be of particular relevance for those making reproductive choices and concerned relatives of an affected family member; beyond confirmation of diagnosis however, it is argued to be of questionable benefit as current testing methods are frequently not able to establish the genetic aetiology, are expensive and, ultimately, do not impact on treatment decisions (McCorquodale, Pucillo and Johnson, 2016).

1.1.5 Treatment. Despite recent clinical trials with promising findings in animal models (Zhao et al., 2017), currently there is no known cure for CMT. Treatment therefore involves management strategies aimed at reducing symptom burden, slowing progression and increasing functional capacity. This can include physiotherapy,
corrective orthotics and splints, pain management medication, and/or surgical intervention (McCorquodale, Pucillo and Johnson, 2016; Rossor, Evans and Reilly, 2015; Yagerman et al., 2012; Pareyson and Marchesi, 2009; Jani-Acsadi, Krajewski and Shy, 2008). Recently, the importance of the psychological experience of CMT has also been recognised, with calls for a multi-disciplinary approach to management of the condition (McCorquodale, Pucillo and Johnson, 2016).

1.1.6 Psychosocial impact. CMT can markedly affect psychosocial functioning (Arnold, McEntagart and Younger, 2005). A recent systematic review found that individuals with CMT have an increased risk of depression and sleep disorders, and generally experience reduced quality of life (Cordeiro et al., 2014). Indeed numerous studies have reported that CMT is associated with reduced health-related quality of life (Calvert et al., 2013; Taniguchi et al., 2013; Boentert et al., 2010; Redmond, Burns and Ouvrier, 2008; Padua et al., 2006, 2008; Vinci et al., 2005; Teunissen et al., 2003).

1.2 Quality of Life

1.2.1 Definitions: Quality of life and health-related quality of life. The World Health Organisation (WHO) defines quality of life as an “individuals' perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns” (WHOQOL Group, 1995, p. 1405). It is suggested that this broad construct can be affected by the individual’s
physical and psychological health, their autonomy, and relationships with others and
the environment (WHOQOL Group, 1995). Health-related quality of life (HRQoL) is
broadly defined as “quality of life relative to one’s health or disease status” (Bakas et
al., 2012, p. 1). It is associated with “optimal levels of mental, physical, role (e.g. work,
parent, carer) and social functioning, including relationships, and perceptions of health,
fitness, life satisfaction and well-being. It should also include some assessment of the
patient’s level of satisfaction with treatment, outcome and health status and with
future prospects” (Bowling, 1995, p.6). A contested term, HRQoL is broadly agreed to
be multi-dimensional, subjective and dynamic (Bakas et al., 2012).

According to Wilson and Cleary’s (1995) conceptual model, health-related
quality of life includes five core components: biological and physiological variables,
symptom status, functional status, general health perceptions and overall quality of
life. These components are argued to be causally linked and influenced by
characteristics of the individual (e.g. personality, motivation, values and emotional
factors) and the environment (e.g. psychological, social and economic support)
(Ferrans et al., 2005; Wilson and Cleary, 1995). Despite the fact this model is
commonly cited within the literature, there is considerable variation in the
conceptualisation and measurement of health-related quality of life in research and
practice (Bakas et al., 2012; Wilson and Cleary, 1995).
1.2.2 The dynamics of health-related quality of life. Health-related quality of life is not a static construct. According to the ‘response-shift’ model (Sprangers and Schwartz, 1999), individuals with a chronic disease must find ways to accommodate their condition. This process of adaption is mediated, in part, by a change in conceptualisation of quality of life, values or internal standards, otherwise known as a response shift. A dynamic feedback loop is suggested to enable individuals to maintain or improve their perceived quality of life (Sprangers and Schwartz, 1999). This may explain why individuals with severe and disabling health conditions may report higher subjective quality of life than their healthy counterparts, a phenomenon known as the ‘disability paradox’ (Albrecht and Devlieger, 1999). The concept of a response-shift also features within the Disability Centrality model (Bishop, 2005). This suggests that following the onset of chronic disease there is an initial reduction in quality of life which can potentially be reversed by a change in perceived importance of affected domains or a change in perceived control. Without such change in perceived importance or control, quality of life may remain impaired (Bishop, 2005).

1.2.3 Assessment and measurement. Measures of health-related quality of life are diverse and include objective and subjective outcomes, thereby reflecting the broad conceptualisation of this construct. One category of measurement which is increasingly common is known as ‘Patient Reported Outcome Measures’ (PROMs) (Neale and Strang, 2015). PROMs can be generic or disease specific, in addition to
domain specific, individualised/ preference-based and utility measures. Although

generic measures (e.g. the SF-36 (Ware and Sherbourne, 2012) are the most frequently
utilised in research, there has been a surge in development of disease specific
measures which may have greater utility within clinical research due to their increased
specificity and responsiveness to change (Garratt et al., 2002). Indeed the development
of measures to assess HRQoL in individuals with CMT has been repeatedly called for,
with clinicians and researchers arguing that multi-disciplinary input will be essential in
their conceptualisation and development (McCorquodale, Pucillo and Johnson, 2016;
Shy and Rose, 2005). Development of such measures is a complex process however,
with a constantly evolving methodology. A typical first stage involves the development
and piloting of a focus group topic guide, informed by the literature and a patient
reference group (Rose et al., 2011). Therefore, this research aims to develop
recommendations for future CMT-specific HRQoL measures through discovery and
explanation of potentially novel domains of HRQoL.

1.3 Charcot-Marie-Tooth Disease and Health-Related Quality of Life

There is widespread agreement that, irrespective of type and severity, CMT
impairs HRQoL (Cordeiro et al., 2014; Shy and Rose, 2005). This is comparable with,
and in some areas, in excess of, other chronic health conditions, such as diabetes
mellitus and epilepsy (Redmond, Burns and Ouvrier, 2008). Understanding the factors
which influence HRQoL in individuals with CMT is crucial to identifying appropriate
interventions to enhance it. Furthermore, measures of HRQoL will be vital for future clinical treatment trials to ensure benefits are meaningful to patients (Shy and Rose, 2005).

1.3.1 Quantitative findings. The current understanding of factors which influence health-related quality of life for individuals with CMT is limited. There is disagreement within the literature as to whether HRQoL is particularly impaired in women (Vinci et al., 2005) or is equivalent across genders (Taniguchi et al., 2013). Moreover, whilst some quantitative studies report largest deficits in social and emotional aspects of HRQoL (e.g. Taniguchi et al., 2013), others report strongest deficits in physical components (Johnson et al., 2014; Redmond, Burns and Ouvrier, 2008).

A range of physical factors have been found to predict HRQoL in individuals with CMT including fatigue, restless leg syndrome and sleep quality (Boentert et al., 2010), lower limb weakness and leg cramps (Redmond, Burns and Ouvrier, 2008), disability (Padua et al., 2006), strength of hand/forearm, walking difficulties and sensory function (Padua et al., 2008) and leg strength (Roberts-Clarke et al., 2016). Thus, to date, there has been a focus on the impact of physical factors, with relatively little attention paid to emotional or psychological factors which are important aspects of HRQoL (Bowling, 1995).

Social and emotional predictors have been largely overlooked in previous quantitative research. Links have been found with depression (Padua et al., 2006) and
employment (Johnson et al., 2014; Vinci et al., 2005). However, not all studies have found occupational status to be a significant predictor of HRQoL (Taniguchi et al., 2013) and research has indicated adults with CMT may not experience any increased risk of depression (Vinci et al., 2009). Thus, current understanding of the social and emotional factors which influence HRQoL is particularly limited and contradictory. This is perhaps indicative of the complex nature of CMT and highlights the need to further explore patient views and experiences.

Given the importance of physical factors assumed by most previous studies, researchers have reasoned that disease progression may also impair quality of life (Cordeiro et al., 2014). However, whilst some studies report a decline in health-related quality of life with increasing age (Johnson et al., 2014; Redmond, Burns and Ouvrier, 2008; Vinci et al., 2005), others find no link with age or disease duration (Taniguchi et al., 2013; Redmond, Burns and Ouvrier, 2008; Padua et al., 2006). One possible explanation for such discrepant findings is the role of acceptance, which is suggested to develop over time as the condition progresses (Cordeiro et al., 2014; Vinci et al., 2005). Acceptance and other positive psychological variables have, however, been poorly researched in individuals with CMT thus far, so such suggestions are speculative. Another possible explanation of the conflicting findings relates to issues with measurement of HRQoL (Cordeiro et al., 2014).

The current confusion surrounding HRQoL and CMT may be attributable to the use of the generic patient-reported outcome SF-36 scale (Ware and Sherbournue, 2012)
to measure health-related quality of life in most previous studies (Taniguchi et al., 2013; Redmond, Burns and Ouvrier, 2008; Padua et al., 2008, 2006; Vinci et al., 2005). Concerns have been raised over the suitability of this scale for patients with neuromuscular disorders, with condition specific HRQoL measures argued to be preferable (Burns et al., 2012). In the absence of a validated CMT specific measure, a qualitative exploration of HRQoL in CMT patients could enable a better understanding of CMT patients’ perceptions of HRQoL, and important influencing factors. This research will begin to address this gap.

1.3.2 Qualitative findings. To date, a small number of qualitative studies have explored the experience of living with CMT using interview or survey methods (Lafarge et al., 2014; Johnson et al. 2013; Ramdharry et al., 2012; Alberts, 2008; Arnold, McEntagart and Younger, 2005). These studies have identified some themes in this experience such as physical symptomology, social and emotional implications, the invisible nature of the condition and resilience (Lafarge et al., 2014). A number of theoretical and methodological limitations to previous research are however evident, such as data collection from a single open-ended survey question (Lafarge et al., 2014) and unspecified analytic frameworks (Ramdharry et al., 2012; Arnold, McEntagart and Younger, 2005; Beyer and Daino, 1991). Previous qualitative research has also been partly deductive, with a pre-determined focus on issues such as fatigue and genetic counselling (Ramdharry et al., 2012; Arnold, McEntagart and Younger, 2005) which
may lead to important insights being overlooked by researchers. Only one previous qualitative study has directly explored CMT patients’ perceptions of factors affecting HRQoL (Johnson et al., 2013). This study is limited however by a focus on symptoms and pre-imposed suggestions of factors likely to affect HRQoL within the interview questions (E.g. “Is excessive sleepiness or fatigue a problem for you?”), which may have influenced responding (Johnson et al., 2013). Furthermore, it is limited by the fact that data were collected using only one mode of enquiry (verbal descriptions provided in an interview) at a single point in time.

HRQoL is a dynamic construct which is likely to vary over time in individuals with CMT (Shy and Rose, 2005). As such, one off qualitative interviews or surveys may not capture HRQoL in appropriate depth and detail. Moreover, qualitative interviews necessarily follow the researchers’ own agenda and arguably produce contrived data (Brunsden and Goatcher, 2007). Recently, qualitative researchers’ over-reliance on verbal descriptions of experience has been criticised. Since different modalities (e.g. verbal, visual, touch etc.) are complicit in an individual’s experience, it has been argued that qualitative researchers seeking to understand experience should adopt a multi-modal approach (Reavey and Prosser, 2012). Thus, researchers have recently advocated the use of novel photo-methods (Williams, Sheffield and Knibb, 2016).
1.4 Photovoice

1.4.1 Definition and approaches. Various photo-methods exist including (1) photo-documentation (documenting visual images related to social processes); (2) photo-elicitation (using researcher or participant generated photos within research interviews); and (3) photo novella or photovoice (a participatory method in which participants choose aspects of their experience to photograph such that the images and their descriptions can then be analysed) (Rose et al., 2011; Berman et al., 2001). Originally developed by Wang and colleagues, photovoice is a method of data collection in which participants are asked to photograph experiences in relation to their individual situation, to represent and enhance their community (Wang and Burris, 1997). Since its initial inception and origins within sociology and community action, the photovoice method has evolved. Indeed, researchers have argued that photovoice can (and should) be adapted to better fit the aims of psychological research (Brunsden and Goatcher, 2007).

Typically, photographs are taken over a period of time and are accompanied by interviews/ focus groups or reflective diary entries to enable the collection of rich data, informed by the participants’ perspective, as opposed to the researchers (Brunsden and Goatcher, 2007; Aubeeluck and Buchanan, 2006). It has been argued that this method enables participants to express meaningful thoughts and emotions in tangible terms, whilst also capturing the temporal nature of experience and facilitating communication and interpretation between researcher and participant. This can lead
to new insights which the participant may not otherwise be able to express (Berman et al., 2001) and may not be captured by one-off researcher-led interviews. Moreover, participant-generated photographs can be helpful in uncovering everyday aspects of experience which may not be explicitly captured via other methods and can be empowering for participants, placing them at the centre of the research process (Rose, 2016).

There is debate in the literature about the most appropriate method to facilitate the ‘voice’ element of photovoice (Topcu, 2015). Although traditionally, focus groups and interviews have been used (e.g. see Frith and Harcourt, 2007; Wang and Burris, 1997), recent evidence suggests accompanying written narratives or diaries are also beneficial. Written narratives reduce the need for researcher interpretation of photos and may therefore increase the validity of the data (Williams, Sheffield and Knibb, 2016). Moreover, written narratives may be preferable to follow-up interviews or focus groups which can be particularly burdensome to individuals with physical impairments (Baker and Wang, 2006). Furthermore, written narratives enable participants to reflect on the meaning of photographs as they are taken; this may be forgotten when participants are interviewed at a later date (Thompson et al., 2008). Written narratives or diaries can help researchers to unobtrusively access accounts of experiences over time and may provide greater depth and understanding than other methods (Hays and Singh, 2011). Additionally, they can be shared with the researcher in a variety of ways (Lyons, Rohleder and Lyons, 2014).
Usually, photovoice studies provide participants with disposable cameras and instructions for use (Wang and Burris, 1994). Smartphones and camera phones are now pervasive within society, however, and are increasingly utilised in health research (Boulos et al., 2011; Kindberg et al., 2005). Indeed, the use of online methods and smartphone applications (apps) has been advocated to facilitate qualitative health research (Garcia, Welford and Smith, 2015). The use of digital photography within photovoice studies has been recently advocated, to reduce the burden of participation (e.g. problems associated with remembering to carry a disposable camera, having to post it back to the researcher etc.) and associated high levels of attrition (Williams, Sheffield and Knibb, 2016; Topcu, 2015). The use of digital photography arguably provides participants with more control over the images they take and enables participants to use online methods to communicate with the researcher about the photographs. Apps in particular provide participants with a mobile way to record experiences in real-time. They also enable researchers to instantly access and monitor data and communicate with participants. Furthermore, apps may be particularly beneficial for sensitive research since they enable inconspicuous participation (Garcia, Welford and Smith, 2015). Although the use of smartphones has recently been advocated for use within photovoice studies (Williams, Sheffield and Knibb, 2016), to date research has not fully utilised this technology.

1.4.2 Limitations of photovoice and smartphone research. Photovoice is not without limitations. The process of taking photos and providing the researcher with accompanying narratives is time consuming and potentially burdensome (Topcu,
Indeed, previous health psychology related photovoice studies have reported high levels of attrition amongst recruited participants (Williams, Sheffield and Knibb, 2016), with typically small final sample sizes ranging from 5-15 (Frith and Harcourt, 2007; Aubeeluck and Buchanan, 2006). Whilst the use of disposable cameras in previous studies has arguably inflated participant burden and associated attrition (e.g. problems with remembering to take the camera out) (Williams, Sheffield and Knibb, 2016), it is not the only source of burden. Moreover, participant burden may be especially pertinent to individuals with a chronic health condition, who may experience fatigue and distress or have difficulties operating a camera and/or accessing desired locations to take photos (Topcu, 2015; Lal, Jarus and Suto, 2012). Thus, researchers should endeavour to reduce burden where possible and conduct research sensitively, providing individuals with some control and flexibility over the pictures they take (e.g. number and frequency) and allowing for others to take photos under the participants’ direction where necessary (Lal, Jarus and Suto, 2012; Frith and Harcourt, 2007).

Despite the advantages of smart phone research, smart phone applications also have a number of limitations. Whilst recent evidence suggests 4 out of 5 UK adults now own a smart phone (Deloitte, 2016), this means that 20% of people do not own a smart phone. Moreover, research has previously found that smart phone ownership is affected by socio-demographic (Pew Research, 2016) and personality factors (Lane and Manner, 2011). Thus, using smart phones to facilitate enhanced access to research data may simultaneously restrict access to participation, thereby limiting the research due to sample biases (Garcia, Welford and Smith, 2015). Additional difficulties with
using apps in research include the challenge of ensuring practical usability of the app, the difficulties in providing clear instructions for use to participants who may be taking part remotely, technological limitations and cost implications. Hence, piloting of the app and alternative options for participation should be considered in smart phone qualitative research (Garcia, Welford and Smith, 2015).

1.4.3 Applications of photovoice. To date, photovoice has been used extensively to capture a range of issues and experiences, often with marginalised populations groups. Topics of study have included, for example, homelessness, poverty, unemployment, and mental and physical health (Cabassa et al., 2013; Walker and Early, 2010; Baker and Wang, 2006; Hergenrather, Rhodes and Clark, 2006; Wang, Cash and Powers, 2000). Researchers have argued that photovoice can capture dynamics of health-related experiences which are not accessible with quantitative methods (Baker and Wang, 2006). Due to the control afforded to participants in how they convey their experiences, photovoice is suggested to be a particularly suitable method for sensitive areas of health research, such as quality of life (Brunsden and Goatcher, 2007).

1.4.4 Photovoice to explore quality of life and HRQoL. A small number of studies have used this method to uncover detailed accounts of quality of life and HRQoL in individuals with chronic illnesses and their families, for example women with
polycystic ovary syndrome (PCOS) (Williams, Sheffield and Knibb, 2016), African American survivors of breast cancer (López and Randall-David, 2005) and carers of individuals with Huntington’s disease (Aubeeluck and Buchanan, 2006). This research has led to the discovery of novel domains of QoL and HRQoL, not identified in previous qualitative studies. For example, one study identified the importance of relationships and connection, including connections with pets, which had not been recognised within previous PCOS research (Williams, Sheffield and Knibb, 2016). Another study identified key social forces (racism, cancer stigma and cultural beliefs) affecting quality of life in African American breast cancer survivors, thus highlighting the need to intervene at different levels (López and Randall-David, 2005). A further study identified themes of ‘care and security’ and ‘small pleasures’ as important to carers of individuals with Huntington’s Disease. Again, these domains of quality of life had been largely overlooked by previous research (Aubeeluck and Buchanan, 2006). Thus, photovoice may enable the identification of new and transformational insights into HRQoL, as it is experienced by adults living with CMT.

1.5 Study Rationale

As previously presented, CMT is a progressive and debilitating neuromuscular condition which adversely affects HRQoL. In the absence of an effective treatment for CMT, preserving HRQoL is of vital importance (McCorquodale, Pucillo and Johnson, 2016). Current validated measures used to assess HRQoL are not condition specific however, and therefore lack relevance to individuals with CMT (McCorquodale, Pucillo
and Johnson, 2016; Burns et al., 2012). Understanding the factors which influence HRQoL for individuals with CMT is important to enable appropriate interventions and development of condition specific measures which could be used in future treatment trials (Shy and Rose, 2005). With its emphasis on participant-generated visual and verbal data, photovoice (Wang and Burris, 1997) can help researchers identify meanings which may not otherwise be accessible. The use of digital photos also enables photovoice to be set-up and delivered online, which may be preferable for individuals with limited mobility. To date, no published research has examined HRQoL in adults with CMT using photovoice, thus findings will make a unique contribution to current knowledge. Findings could have important implications for the understanding of HRQoL in adults with CMT and inform recommendations for related assessment and intervention. Furthermore, no published photovoice study has used an online version of photovoice, thus the current study uses a novel approach. Consequently, findings from the current study could also inform developments of the photovoice method within psychological research.
1.6. Aims and Objectives

In light of the background literature and project rationale, this research will have the following overall aim and objectives:

**Aim:**

- To explore how adults living with CMT understand HRQoL and the factors which influence this.

**Objectives:**

- To describe, in detail, HRQoL and the factors which influence it using photographs and reflective diary entries generated by adults living with CMT.
- To access every-day experiences by gathering participant-generated photos and verbal interpretations online, over a two-week period.
- To develop recommendations for future CMT-specific HRQoL measures through discovery and explanation of potentially novel domains of HRQoL.
- To identify potential interventions to enhance HRQoL in adults with CMT.
2. Methodology

2.1 Philosophical Paradigm, Ontology and Epistemology

A pragmatist perspective underpins this study. Pragmatism rejects the realism – constructionism debate and focuses instead on the purpose and consequences of knowledge. Originally outlined by American philosophers such as John Dewey, William James and George Herbert Mead, pragmatism has recently been advocated as a useful paradigm to address “the problem of knowledge in health psychology” (Cornish and Gillespie, 2009, p.3). Pragmatism accepts the existence of various forms of knowledge and competing interests (pluralism), however it argues that such knowledge can be evaluated with respect to successful action (non-relativist). Moreover, pragmatism questions whose interests are served by knowledge and prioritises everyday problems and actions (action-orientated) as the primary reality and test of knowledge (Cornish and Gillespie, 2009). From a pragmatist perspective, ‘truth’ is simply “what works at the time” (Creswell, 2014, p. 10).

Beliefs about the nature of reality are known as ontology. Various ontological perspectives exist ranging from realist (objective reality or ‘truth’ exists) to relativist (reality is constructed and co-constructed) (Killam, 2013; Lincoln, Lynham and Guba, 2011). From a pragmatist perspective, questions about the nature of reality are redundant (Deforge and Shaw, 2012; Cornish and Gillespie, 2009). Whilst pragmatists
generally acknowledge the existence of an external reality, it is argued that knowledge of this can never be fully attained and therefore debating the nature of reality is of limited value (Deforge and Shaw, 2012). Instead, knowledge is seen as a tool for action that helps to solve problems. For pragmatists, meaning is found within the transformation of experience through inquiry. Indeed experience (present and future) is considered a key ontological category for pragmatists (Rosiek, 2013). The value of knowledge therefore lies not within its representativeness of reality, but rather in its usefulness for a given interest or purpose (Cornish and Gillespie, 2009). Thus, pragmatism is associated with an “ontology of the future” (Rosiek, 2013 p.692).

Ontological perspectives necessarily influence epistemology. In essence, epistemology refers to beliefs about the way in which we derive knowledge. This includes the relationship between knowledge and the researcher (Killam, 2013). Pragmatists recognise that research occurs within a broad context, including historical, social and political factors (Creswell, 2014). From a pragmatist standpoint, there is no preferential method of establishing knowledge. Instead, the ‘best’ method is the one which achieves its purpose in addressing the research question (Cornish and Gillespie, 2009).
2.2 Methodological Theory

In order to appropriately address the current research questions, this research adopted an inductive approach whereby meaning was generated from the data. This contrasts with the deductive approach in which theory is tested (Creswell, 2014). In accordance with the inductive approach, no theories were tested in this research. However, the underlying premise for the research was generally informed by Wilson and Cleary’s (1995) conceptual model of HRQoL, the ‘response-shift’ model (Sprangers and Schwartz, 1999) and the Disability Centrality model (Bishop, 2005), which collectively emphasise the complex and individual nature of HRQoL.

2.3 Methodology

Photovoice is an approach in which participants are asked to photograph aspects of their everyday experiences over a period of time (Wang and Burris, 1997). Originally outlined by Wang and Burris (1994), the ‘voice’ element of ‘photovoice’ is described as “Voicing Our Individual and Collective Experiences” (p. 381). Unlike other visual methods such as photo-elicitation, participant-led photo generation is central to the photovoice research method (Brunsden and Goatcher, 2007). Given the multi-modal nature of human experience, reliance on verbal descriptions alone may overlook important aspects of experience (Reavey and Prosser, 2012), thus photovoice has the potential to identify new understandings and insight. Since its initial inception as a method for participatory and community action research (Wang and Burris, 1997),
photovoice is argued to be a flexible and adaptable method which can readily be applied to psychological research (Teti et al., 2016; Brunsden and Goatcher, 2007). Furthermore, as photovoice is particularly useful for uncovering rich and detailed accounts of experience, it is increasingly utilised within health psychology research (Topcu, 2015). Indeed, researchers have argued that photovoice can capture dynamics of health-related experiences which are not accessible with other methods (Baker and Wang, 2006). Furthermore, advances in digital technology offer a new way of conducting photovoice investigations which may reduce participant burden and attrition (Williams, Sheffield and Knibb, 2016; Topcu, 2015).

From a pragmatist perspective, photovoice can aid in understanding commonalities and differences in experiences in order to contribute to theory and practice (Hansen-Ketchum and Myrick, 2008). Provided there is internal (within participant) and external (between participants) replication of findings, it can also provide a reliable representation of the priorities of individuals, at a particular point in time (Wang and Burris, 1997). The ultimate aim is, however, not to provide an objective description of experience but instead, to provide a useful interpretation to aid desired action (Rosiek, 2013; Cornish and Gillespie, 2009). As a tool for psychological research, the aims of photovoice are to provide visual data relevant to the concerns of a particular population group, to provide verbal descriptions of the photographs, and to contribute to psychological theory and practice via combined analysis of visual and verbal data (Brunsden and Goatcher, 2007).
2.3.1 Identifying an analytic strategy. Photovoice enables the analysis of both visual and verbal data. The analytic strategy adopted in previous research is diverse, ranging from content analysis (Aubeeluck and Buchanan, 2006), to thematic analysis and iconography (Mizock, Russinova and Shani, 2014) and grounded theory (López and Randall-David, 2005). It is often unclear, however, whether photographs have been analysed or are simply used to illustrate themes derived from analysis of the verbal data alone (Brunsden and Goatcher, 2007). The latter strategy is problematic since potentially meaningful data is not utilised (Brunsden and Goatcher, 2007). Indeed research has found that photographs can convey meaning in the absence of textual explanation (Byrne, 2014). Methods of analysis for visual data are, however, often unclear and greater clarity in reporting of analytic strategies within visual research has been repeatedly called for (Byrne, 2014; Brunsden and Goatcher, 2007). Typically, some form of categorisation (e.g. via content analysis) is utilised but approaches to this vary and lack detail (Byrne, Daykin and Coad, 2016). Whilst specific visual methods such as semiology and iconography exist, these are concerned with cultural meaning and compositional and social modalities respectively. They are less helpful in addressing affective (semiology) and lived (iconography) experiences (Rose, 2016). Recently, psychologists have begun to outline new analytic strategies for visual data linked to thematic analysis, such as ‘polytextual thematic analysis’ (Gleeson, 2012) and ‘thematic visual analysis’ (Byrne, 2014). In polytextual thematic analysis, repeated viewing of images is followed by noting down initial thoughts and feelings to generate
proto-themes which are checked for distinctiveness and refined as necessary (Gleeson, 2012). The analysis for this study was informed by this approach, in addition to general guidelines for thematic analysis (Braun, Clarke and Terry, 2015). Thematic analysis is preferable to other methods such as IPA which necessitates an idiographic focus, and is most suited to qualitative interviews (Smith, 2008).

Whilst some researchers argue that computer assisted qualitative data analysis can restrict the analytic process (Blismas and Dainty, 2003), others dispute this and emphasise that NVivo improves the robustness and transparency of qualitative analysis (Bergin, 2011). NVivo was chosen for the current study because it can be used to facilitate the storage and analysis of visual and textual data in combination.

2.4 Methods

2.4.1 Design. In keeping with the pragmatist assertion that multiple methods can be utilised effectively within research (Creswell, 2014), a multi-modal qualitative design was adopted. This involved a photovoice study which incorporated participant-generated photographs and accompanying written narratives. Qualitative researchers’ over-reliance on verbal descriptions of experience has recently been criticised. Since different modalities (e.g. verbal, visual, touch etc.) are complicit in an individual’s experience, it has been argued that qualitative researchers seeking to understand experience should adopt a multi-modal approach (Reavey and Prosser, 2012).
2.4.2 Patient and public involvement. A collaborative approach to patient and public involvement (PPI) (INVOLVE, 2012) informed this study from an early stage. The Chief Operating Officer for the UK-based charity CMT-UK was recruited as a patient partner (see Appendix VIII for patient partner agreement). As part of this role, the patient partner (who also has a diagnosis of CMT) commented on the proposal and the documentation for participants (e.g. participant information sheet), tested the online consent and details form and study website/ app and advertised the study to participants. After the study was completed, the patient partner also commented on a draft of the analysis chapter and endorsed the findings. Collaborative approaches to PPI help to ensure research is relevant and ethical and can help with recruitment (INVOLVE, 2012). Due to the geographical distance between the patient partner and the researcher, and the lack of available funding to reimburse costs, communication took place via email.

2.4.3 Sample and recruitment. Adults living with CMT were recruited online via a self-selecting sample. The study was advertised by CMT-UK, via their social networking sites and newsletters. Additional CMT and neuromuscular charities were added as a back-up recruitment strategy via an ethical approval amendment. However, all participants in the present study were recruited via CMT-UK. The CMT-UK Facebook page is followed by 3,000+ people, including members and non-members. This
strategy was chosen as a pragmatic approach to recruiting individuals with a rare condition. The aim of this study was not to develop a representative sample but rather to create a framework of understanding.

Adults with CMT who expressed an interest in the study advert were emailed with the participant information sheet (see Appendix B II), a photovoice information sheet (see Appendix B V) and a web link to the password protected online consent and participant details form (hosted by Qualtrics1) (see Appendix B III - IV).

The following inclusion and exclusion criteria were applied:

Inclusion criteria: Male and female adults (18+) with a medical diagnosis of CMT (any type) were eligible to participate. In addition, due to the method, access to the internet and the means to take digital photographs (e.g. via a smart phone or digital camera) were also required.

Exclusion criteria: As this was not a funded project, access to translation services for non-English speaking participants was not possible. Therefore, individuals fitting the above criteria who do not speak English were not eligible to participate. Furthermore, individuals with specific co-morbid conditions which could cause significant cognitive impairment and therefore affect mental capacity were not eligible to participate (e.g.

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1 [https://qualtrics.com](https://qualtrics.com) Link to online consent and participant details form: [https://bristol.az1.qualtrics.com/jfe/form/SV_bdsI9UXKW1CR9Ot](https://bristol.az1.qualtrics.com/jfe/form/SV_bdsI9UXKW1CR9Ot) (password = photovoice)
dementia, brain injury, severe learning disability, severe mental health condition such as schizophrenia).

2.4.4 Method of data collection. The current study utilised an online photovoice journal. This was hosted by Blackboard Learn at the University of Worcester\(^2\) and available to participants via smart phone application or webpage alternative. The online journal was tested extensively by the research team, including the patient partner, to check for functionality. The journal was set up such that participants could only see their own entries. Participants who consented to take part were contacted by email with written and visual information about accessing the journal and provided with an individual, anonymised log-in (see Appendix B VI for information sheet and video links provided to participants about accessing and using the journal). Participants were instructed to take digital photographs (e.g. using a smart phone and/or digital camera) of anything which they felt affected their quality of life (positively, negatively or neutral) over a 2-week period. This period was chosen as an appropriate time frame within which to capture dynamic experience (Topcu, 2015). In line with previous similar research, no definition of HRQoL was provided to participants since the research aimed to examine how participants understood HRQoL and the factors that influence it (Williams, Sheffield and Knibb, 2016). Whilst the participant information sheet did not specify the term HRQoL, participants were primed with the

\(^2\) [https://worcesterbb.blackboard.com](https://worcesterbb.blackboard.com)
sentence “Research suggests that living with Charcot-Marie Tooth (CMT) disease can affect an individual’s quality of life.” Thus, it was assumed participants would interpret this as HRQoL. Participants were also asked to produce associated reflective diary entries explaining what each photograph meant, why it was important to the participant and how what it depicted influenced their quality of life. In accordance with previous research, flexibility in the number and frequency of entries was encouraged (Williams, Sheffield and Knibb, 2016), although a suggested limit of 25 combined photograph and diary entries was advised. To reassure participants that their posts were being received, and to promote engagement, standardised comments were provided after the first journal entry, at the end of week 1 and at the end of week 2. Given the novel aspects of the above method, the procedure was initially tested with two participants to establish feasibility before the remaining participants took part. Following the initial pilot, minor changes were made to researcher-participant communication strategies (e.g. participant confirmation of successful first post via email instead of within the journal). No changes were made to other aspects of the study design and therefore data from the first two participants is included within the overall analysis.

2.4.5 Method of analysis. The analysis was conducted according to the following steps:
1. Data storage - Each individual journal entry was transferred verbatim into NVivo 9, with photographs and accompanying textual reflections stored together within sources. Sources were labelled according to participant numbers and were grouped by participants.

2. Familiarisation with the data – this began with reading/re-reading and viewing/re-viewing the written text and images, in different orders. Initial reflections on meaning, immediate emotional responses and similarities and differences between images and texts were noted and stored as annotations within NVivo. After this, a written description and interpretation of each image was added beneath the participant’s own narrative, linked to the image within NVivo. Different text colours were used to demarcate description or interpretation linked to the participants’ reflective text (pink) and that which originated from the researcher (purple).

3. Coding the data - To ensure consistency in the analysis of the visual and verbal data, inductive coding was conducted on both the images and accompanying textual reflections. During the coding process, the image(s), participants’ accompanying text and the researcher’s description/interpretation of the image(s) were all visible on the screen within each individual journal entry. Coding was conducted inductively, in order of journal entry. Coded extracts of reflective text, photographs and accompanying description/interpretation were collated for each code. Codes were checked for consistency and refined,
to ensure patterns of meaning were captured effectively across diverse perspectives (Braun and Clarke, 2012).

4. Searching for themes – Similar codes and clusters of meaning across the data were subsequently grouped together to develop the main themes. During this process, the central organising concept of each theme was identified to check for theme coherence and distinctiveness (Braun, Clarke and Terry, 2015).

5. Review themes – Themes were discussed in supervision and checked against the visual and verbal data. A thematic map was subsequently generated to determine inter-relationships between the themes and an overall structure for the analysis (Braun, Clarke and Terry, 2015).

6. Defining themes – Theme definitions and titles were developed and refined to ensure they appropriately captured meaning in each theme. Extracts of text and photographs which best illustrated each theme were subsequently identified for inclusion in the written analysis.

Thematic analysis is not a mechanical, linear process but instead is flexible and organic (Braun, Clarke and Terry, 2015). As such, researcher reflexivity is essential and will be discussed separately below. Furthermore, the analysis often involves a degree of moving forwards and backwards as insight develops. This was the case in the present study. Adjustments to the analysis were made at every stage and the write-up itself was crucial in refining the analytic narrative.


**2.4.6 Ensuring Quality and Rigour.** Various guidelines exist for establishing quality in qualitative research (e.g. Tracy, 2010; Meyrick, 2006; Yarley, 2000). The eight “big-tent” criteria for excellent qualitative research guided and informed this study (Tracy, 2010). These criteria are designed to flexible across qualitative methods and their respective philosophical underpinnings. They include: worthy topic, rich rigour, sincerity, credibility, resonance, significant contribution, ethical and meaningful coherence (Tracy, 2010).

In addition to addressing a significant and important issue, this study used a novel research method and thus can be considered a worthy topic which makes a significant contribution to the field. Data collection and analysis were conducted rigorously, with rich descriptions provided to illustrate key themes and attention directed towards ensuring “empathic validity” (Dadds, 2008) and resonance of the study, in addition to overall meaningful coherence (Tracy, 2010). Quality issues pertaining to credibility, sincerity and ethical issues will be discussed in more detail below.

To ensure ‘credibility’ in the current study, crystallization was utilised. Crystallization refers to the process of gathering multiple types of data and involving multiple researchers/ viewpoints to develop a deeper understanding of an issue. It differs from triangulation which seeks to establish a singular truth or reality via consensus (Tracy, 2010). Accordingly, the analysis was conducted on the photographs and textual reflections (multiple sources of data). Furthermore, coding of a complete
set of journal entries for one participant was conducted by the researcher and lead supervisor to facilitate a discussion around interpretations. Additionally, initial themes were discussed and interrogated within the supervisory team. The patient partner for the study also provided a reflective commentary on the draft analysis (multiple viewpoints and researchers). Inter-rater reliability was not calculated since this is based on the premise that an accurate reality exists which can be captured by coders (Morse, 1997). This is not consistent with pragmatist assumptions which emphasise the usefulness of findings, rather than their correspondence to an external reality (Deforge and Shaw, 2012). Furthermore, inter-rater reliability is not appropriate for unstructured approaches in qualitative methods and is likely to generate superficial coding which does not adequately capture the richness and depth of the data (Morse, 1997).

2.4.7 Reflexivity. According to Tracy (2010), a key component of ‘sincerity’ in qualitative research is self-reflexivity. Reflexivity refers to the process of critical evaluation of the researcher’s position in relation to the research, and the impact of this position on the research process and outcome (Berger, 2013). Reflexivity is important within pragmatic enquiry generally (Rosiek, 2013) and within qualitative research methods specifically (Yarley, 2000). In addition to general positioning of the researcher with respect to their personal characteristics, beliefs, preferences and
biases, reflexivity is affected by whether the researcher shares the participants’ experience and, as such, is part of the ‘researched’ (Berger, 2013).

As a 31-year-old, white, female, University lecturer, with a diagnosis of CMT since childhood, my reasons for conducting this research were inevitably shaped by my own lived experiences. It is important to acknowledge that my initial awareness of and interest in the topic area arose from having the condition myself. In my experience, many people (including numerous medical professionals) have not heard of CMT and have limited knowledge of its complex impact on HRQoL. This experience led me to review the literature where I noticed that my own observations where indeed matched by a deficit in available research.

Prior to starting the research, I did have some concerns about inadvertently influencing what participants said about their HRQoL and I believe this affected my choice of method. In addition to the well-documented benefits of photovoice (see Topcu, 2015 for a review), I felt the revised online method would enable participants to freely express what they wanted to, without me unintentionally re-enforcing aspects I particularly identified with.

Having shared experience with participants can facilitate recruitment and enables the researcher to approach the study and its analysis with valuable knowledge and insight. However, it can also potentially lead to blurred boundaries, projection of biases, assumed knowledge and a feeling of competition or comparison between the researcher and participants (Berger, 2013). In order to mitigate these risks, it is advised
that researchers keep a reflective log during data collection/analysis, undertake repeated review of data and engage in peer consultation (Berger, 2013). Given my own experience of CMT, I followed these guidelines (e.g. see Appendix C II for example reflective diary entries) and engaged in additional activities to reduce the potential for problems arising from this shared experience. To enhance my understanding of my own positioning, I completed a reflective exercise to ascertain my individual perspective on HRQoL (see Appendix C I). This helped me to identify my beliefs about HRQoL prior to engaging in the analysis. Consequently, I was aware of my own biases during the analytic process. Furthermore, in the limited interaction I had with participants, I did not disclose my own CMT diagnosis. This was not an intentional strategy and is perhaps partly reflective of the lack of face-to-face contact between the participants and myself. Whilst self-disclosure can be beneficial, the potential for (and adverse impact of) assumed knowledge and comparison may have outweighed such benefits in the current study design.

Carrying out qualitative analysis can be an emotive process, irrespective of shared experience (Braun, Clarke and Terry, 2015). During the analysis, I experienced a range of emotional responses, from sadness to inspiration. I ensured I noted these experiences down and discussed them in supervision (see Appendix C II for example reflective diary entries). This helped me to fully engage with the data from a position of self-awareness.
2.5 Ethical considerations

Ethical approval was granted from the Health and Applied Sciences Faculty and Ethics Committee, University of the West of England (UWE) (Approval reference number = HAS.16.10.034) (See Appendix B I). As the researcher was employed by the University of Worcester, ethical approval from UWE was registered with the University of Worcester’s Institute of Health and Society Ethics Committee. The BPS Codes of Human Research Ethics (2014) and Internet-Mediated Research (2017) were adhered to.

2.5.1 Informed consent. In accordance with the above guidelines, participants were fully informed about the nature of the research prior to taking part. Specifically, all participants were informed of the aims of the study, the design, what their participation would involve and how it would contribute to the analysis and project write-up. The participant information sheet (see Appendix B II) and consent form (see Appendix B III) were reviewed by the patient partner to ensure they were clear, accessible and sufficiently informative. Participants were encouraged to take time to carefully consider the information sheet before providing consent (at least 24 hours).
2.5.2 Anonymity and storage of data. Participants’ personal information (e.g. name, age, email address) were stored securely in an encrypted password protected file, on a secure University of Worcester network. A University of Worcester hosted website/app (Blackboard Learn) was utilised to enable the secure transfer of photographs and diary entries from the participant to the researcher. For the purposes of analysis, text entries and associated photographs were downloaded and stored securely in NVivo, within a password protected file. A data management plan and data processing agreement were completed (see Appendix B VII and IX).

2.5.3 Withdrawal from the study. All participants were fully informed of their right to withdraw without providing a reason and with no penalty. Participants were advised that they could withdraw their data up to four weeks after completing the study. This time limit allowed sufficient time for participant reflection whilst also ensuring that data could be feasibly removed from the analysis. Participants were provided with information about how to request withdrawal from the study within the participant information sheet. In this instance, participants were required to email the lead researcher with their website log in details.

2.5.4 Photovoice ethics. Photovoice studies give rise to particular ethical issues which must be appropriately addressed. Recent guidelines for ethical visual research methods identified six categories of ethical issues: confidentiality, minimising harm,
consent, fuzzy boundaries, authorship and ownership and representation and audiences (Cox et al. 2014). To address issues of confidentiality, journal entries (photographs and written narratives) were stored securely according to a data management plan. Participants were advised that quotations and photographs would be anonymised and that photographs of identifiable people would not be included within the write-up and dissemination of the study. To minimise harm, participants were provided with information about legal and ethical aspects of photography, emphasising the importance of safety and responsibility (Wang and Redwood-Jones, 2001). Furthermore, details were provided to participants on sources of help and support in case they felt in any way distressed by the experience of taking photographs and reflecting on their HRQoL. To address issues related to consent, participants were instructed to ask full verbal permission from adults in places where privacy could be reasonably expected and full verbal permission from parents/guardians of children. Information about the project (including the researcher’s contact details) was included in participants’ information sheets to give to prospective subjects or parents/guardians. Participants were fully informed of their right to withdraw and that, in this instance, journal entries would be permanently deleted. In addition, participants had the facility to edit and delete journal entries as they deemed appropriate within their study period. Fuzzy boundaries can occur when participant and researcher roles become blurred (Cox et al. 2014). To address this issue, participant and research team roles were clearly defined from the project outset. Authorship and ownership was also addressed from the outset. Thus, the consent form included consent for the research
team to use and publish photographs (in addition to anonymised quotations) within
the analysis and dissemination of the study. Finally, to address representation the
analysis was conducted sensitively and in consultation with the patient partner.
Photographs have been accompanied by associated excerpts from participant diary
entries to avoid misinterpretation.
3. Findings

3.1 Participant Details and Demographics

Thirty-two adults with CMT responded to the online study invitation requesting further information about the study. Of these, 16 people consented to take part in the study and were provided with a journal log-in and instructions for use. Five people who consented did not access and/or post any journal entries. Thus, the final sample comprised 11 adults, three males (27%) and eight females (73%). None of the participants were known to the researcher. Nine participants were resident in the UK; the remaining two participants were resident in Europe and Canada. The mean age was 55 (range = 27 – 75), with a mean of 22 years since diagnosis (range = 1 – 63). Three participants (27%) were not aware of, or did not provide information about, their type of CMT. Of the participants who provided this information, type of CMT was equally split between Type 1 (4 participants = 36%) and Type 2 (4 participants = 36%). Key demographic details are presented in Table 1.
3.1.1 Table 1 Demographic Data for Individual Participants

<table>
<thead>
<tr>
<th>Pseudonym</th>
<th>Age</th>
<th>Gender</th>
<th>Employment Status</th>
<th>CMT Type</th>
<th>Years Since Diagnosis*</th>
<th>Age at Diagnosis*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Paul</td>
<td>64</td>
<td>Male</td>
<td>Retired</td>
<td>Not answered</td>
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<td>59</td>
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<tr>
<td>John</td>
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<td>Male</td>
<td>PT</td>
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</tr>
<tr>
<td>Millie</td>
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<td>Female</td>
<td>FT</td>
<td>Not answered</td>
<td>5</td>
<td>22</td>
</tr>
<tr>
<td>Sarah</td>
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<td>Female</td>
<td>Retired</td>
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<tr>
<td>Robert</td>
<td>65</td>
<td>Male</td>
<td>Retired</td>
<td>Type 1A</td>
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<td>34</td>
</tr>
<tr>
<td>Jill</td>
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<td>Female</td>
<td>Retired</td>
<td>Type 2A</td>
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<td>28</td>
</tr>
<tr>
<td>Helen</td>
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<td>Retired</td>
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<tr>
<td>Rose</td>
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<td>Female</td>
<td>PT</td>
<td>Type 2</td>
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<td>8</td>
</tr>
<tr>
<td>Jessica</td>
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<td>Female</td>
<td>FT</td>
<td>Type 1A</td>
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<td>38</td>
</tr>
<tr>
<td>Lynn</td>
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<td>Female</td>
<td>PT</td>
<td>Type 1A</td>
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<td>66</td>
</tr>
<tr>
<td>Kate</td>
<td>48</td>
<td>Female</td>
<td>Homemaker</td>
<td>Type 1A</td>
<td>41</td>
<td>7</td>
</tr>
</tbody>
</table>

Note. Pseudonyms have been allocated to protect participants’ identities. PT = Employed/self-employed part-time; FT = Employed/ self-employed full-time

*Diagnosis refers to a formal medical diagnosis of CMT.
3.2 Main Findings

Across the 2-week study period, the mean number of journal entries each participant completed was 8.36 (range = 2 - 14). The mean number of photos each participant uploaded within these entries was 11 (range = 2 – 22). Thematic analysis of the journal entries (photographs and accompanying narratives) identified six main themes. A thematic map representing the relationships between themes and links to HRQoL is presented at the end of the results section in Figure 1. A list of themes and related codes are presented in Table 2. As previously presented, participants were not provided with the specific term ‘HRQoL’. Instead this was primed within the information provided to participants. Thus, references to quality of life were interpreted as HRQoL. All photographs within the analysis are used with the permission of the contributor.

3.2.1 Table 2: Themes and codes from the synthesised thematic analysis of participants’ photographs and written narratives

<table>
<thead>
<tr>
<th>Theme</th>
<th>Sub-theme</th>
<th>Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td>The challenge of the every-day</td>
<td>1. Physical challenges of CMT</td>
<td>Loss of muscle strength, balance, mobility and dexterity; Feet: awkward and hazardous; Chronic pain; Constant fatigue; Stress makes CMT worse; CMT</td>
</tr>
<tr>
<td>Maintaining independence: adaption, adjustment and support</td>
<td>Independence is important; Pro-active and pragmatic adjustment; Adaptions increase independence and reduce risk; The compromise: adaptions are not a perfect solution; Surgery: important benefits but risks and losses; self-management; Car facilitates independence; Independence contingent on disability benefits/ support; Spousal support important for independence; Assistance and support from significant others</td>
<td></td>
</tr>
<tr>
<td>---</td>
<td>---</td>
<td></td>
</tr>
<tr>
<td>2. Navigating restrictive environments</td>
<td>affects everything; Seasonal variation Restrictive environments; Missing out on experiences and disconnection from the norm; Created problems: frustration with others; Everyday activities are frustrating and hazardous</td>
<td></td>
</tr>
<tr>
<td>Lack of control and future-orientated anxiety</td>
<td>Coping self-efficacy; lack of control over assistance;</td>
<td></td>
</tr>
</tbody>
</table>
uncertainty caused by benefit changes; anxiety about progression; The burden of inheritance

<table>
<thead>
<tr>
<th>CMT awareness</th>
<th>Medical professionals are not aware of CMT; Delayed diagnosis; Lack of understanding about CMT; Difficulty disclosing an unknown and unseen condition; Walking aids as communication; Importance of CMT awareness; Support from professionals</th>
</tr>
</thead>
<tbody>
<tr>
<td>Threats to identity and self-esteem</td>
<td>Loss of role and identity; Altered relationships; Appearance concerns; Impact on femininity; Embarrassment</td>
</tr>
<tr>
<td>Resilience and positive growth</td>
<td>Finding positives and meaning; Importance of relationships and connection; Maintaining valued activities and roles; Resilience</td>
</tr>
</tbody>
</table>
3.2.2 Main theme: The challenge of the every-day

This theme relates to the every-day challenges faced by individuals with CMT. It is comprised of two sub-themes: “Physical challenges of CMT” and “Navigating restrictive environments”. Participants described and photographed many debilitating and distressing physical manifestations of CMT. Participants also illustrated and described a range of environments and experiences that are often difficult to access with restricted mobility and dexterity. As a consequence of an interplay between physical manifestations and the external physical and social environment, every-day activities were often described as challenging and frustrating, negatively impacting on HRQoL.

Physical challenges of CMT

All participants illustrated and described the loss of muscle strength, balance, mobility, dexterity and sensation characteristic of CMT. Furthermore, most participants discussed experiencing chronic pain and fatigue. Although participants varied greatly in the extent of their disease progression and functional abilities, all physical manifestations were frustrating and typically had a detrimental impact on perceived HRQoL.

Although Millie was able to walk independently, she described frustration with the shape of her feet:
“One of the main, noticeable things is a high instep and often a turn in the ankle, meaning that your foot never sits flat to the floor or indeed to the inside of your shoes when you're wearing footwear.”

Jill also illustrated the challenges associated with the shape and functionality of her feet:

“This left foot is very deformed and although it spends all day encased in an AFO³ it doesn’t stop the foot being a trip hazard.” (Jill)

The impact of muscle weakness on mobility and HRQoL was likened to the concept of walking with weights by one participant: “Most days at some point you feel

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³ AFO = Ankle foot orthosis. These are braces or orthotics for the ankle and foot to provide stability and support.
like your legs and feet are wearing concrete boots and you just don't feel strong enough to carry yourself around... it effects everything that you do” (Rose). Although a small number of (typically younger) participants were able to walk independently, this was not the case for many participants and, at the extreme, one participant stated: “I can no longer walk, stand or move my feet” (Sarah).

Although mobility-related difficulties were inherently frustrating and distressing, for many participants, loss of dexterity and hand-strength was also challenging. One participant photographed her hand and reflected on her lack of muscle:

“A surgeon who operated on my feet...described my hands as being like a Railway station sandwich "All bread and no filling" to describe the lack of muscle” (Jill)
Other participants elaborated further and discussed loss of hand and wrist function as especially devastating, thus highlighting the differential impact of physical manifestations of CMT:

“Losing the use of my hands has caused more feelings of desperation than losing my ability to walk ever did...Losing the use of your hands, I have found, is worse than losing the use of your legs. Your legs may take you places but your hands make your life for you.” (Sarah)

Most participants identified chronic, unmanageable pain as a common, distressing manifestation of CMT. Three participants dedicated an entire journal entry to pain; such was the enormity of its impact. Rose described the extensive daily pain she experiences:

“One of the biggest impacts on your quality of life with CMT is daily pain. This can range from cramping anywhere in your legs and feet. Sharp stabbing pains, throbbing, constant dull aches, hot pains, pins and needles and just about any other type of pain you can think of. You get pain from walking on deformed feet, pain from nerves not working correctly, aches from muscle fatigue to name but a few.” (Rose)
Sarah uploaded a painting she had painted to represent her pain and what it feels like:

![Painting](image)

In addition to reflecting the fact that “No one can see your pain…” (Sarah), Sarah’s painting conveys the deeply unpleasant and distressing nature of pain. This is further evident in her accompanying narratives: “Pain is stressful because it touches every facet of your life: those you love, your work, play, and sleep, even how you dress.”;

“When you have constant pain the sun doesn’t shine as bright as it used to, music doesn’t delight your soul as it once did and the world isn’t quite as nice a place as it could be.”
The concept of pain and darkness was further elaborated by Helen who chose to photograph the night’s sky. For Helen, night-time pain was relentless and interrupted her sleep:

“And so the pain starts it never fails to sneak up on me every night the same I think oh tonight it will be ok but no always the same. Even after my meds I am awake for hours until the pain gets so bad I sort of just drop off!!! I don't know whether I'm so tired I can't stay awake or my brain says you've had enough go to sleep! Same every night even red wine doesn't help haha” (Helen)

Although there is clearly an intended humour in the idea of red wine for pain management, many participants identified considerable difficulties in managing pain via conventional medication. For Rose, the painkillers depicted in her photo below were a last resort:
“Then of course you also get all the problems associated with taking pain relief on a regular basis. I try to only resort to pain relief when I am absolutely desperate and can’t cope with the pain anymore.” (Rose)

Partly as a consequence of pain, and partly due to muscle weakness and associated problems, most participants described and illustrated constant fatigue, which detrimentally influenced HRQoL. Though not always recognised as part of CMT, fatigue was explained to one participant as a consequence of the fact “…we are trying to do what everybody else does with half the muscle mass” (Sarah).

Sarah photographed herself in bed, illustrating the fact she has to regularly rest, and thus, the debilitating impact of CMT. This was a source of intense vexation for Sarah who commented, “Fatigue is one of the hardest things I have to deal with”. Other participants’ reflections resonated with this sentiment: “CMT can make you almighty tired.” (Lynn).
Although most participants described being physically fatigued from the exertion of every-day life, mental fatigue was also an issue for some:

“Ok so I have made it down the stairs and out of the front door, the amount of concentration to just put one foot in front of the other is tiring. My condition means I have to make sure I fully concentrate on whatever I do which does lead me to become mentally fatigued.” (Robert)

For some participants, the physical manifestations of CMT resulted in, and were made worse by, stress, thereby generating a self-perpetuating vicious cycle. The photo and accompanying narrative below convey a sense of helplessness related to this vicious cycle:
“This photograph signifies stress. With CMT stress has a real negative effect on the condition. Whenever I am stressed I instantly feel a demise in my ability to walk properly. I feel as if the whole of my body is jangling with nerves and I can't seem to function properly. I will drop things, trip up, fall down, walk into things and generally lose control of my peripheries. I have spoken to other members of my family with CMT and we all appear to cope with stress very adversely.” (Rose)

The combination of the various physical manifestations of CMT was considered to be particularly challenging. For example, Sarah commented that the numerous physical consequences she experienced had significantly affected her life:

“Not being able to walk or stand, losing the use of my hands, not being able to breathe very well, having digestion and elimination problems, living with chronic neuropathic pain and muscle spasms, having surgery and continually having to
contend with fatigue and abnormal limbs has factored into my life and changed just about everything I do.” (Sarah)

Navigating restrictive environments

Most participants photographed and wrote about environments and experiences they found difficult to access with reduced strength and mobility. At times, problems with accessibility were exacerbated by the attitudes and actions of others. Every-day activities were often depicted as frustrating and hazardous. This was, in part, a consequence of an interplay between the physical manifestations of CMT and environmental/ societal restrictions.

Paul photographed the outdoor surfaces he finds particularly difficult to negotiate:

Whilst these photos appear innocuous, for those with CMT they represent significant challenges:
“Well as the first photo shows - cobbled streets! Especially if wet and covered in the ‘wrong leaves’! Add a bit of a slope and I might as well give up. Equally tricky are steps without any handrails as shown in the second photo such as football terraces...Going up is not so bad because if I lose my balance at least I can sink to my hands and knees but coming down is a nightmare...” (Paul)

Using stairs with handrails was also frustrating and hazardous for some participants, particularly in less accessible environments. Two participants photographed and wrote about their experiences of using stairs one day:

“The cafe is above a health food shop ... stairs have to be used! I can manage, one at a time but they make life harder. And once upstairs, there is a notice informing that the toilets are downstairs!” (Kate)

“This image shows my stairs to access my flat, they are quite steep. It was starting to get quite difficult to go down the stairs due to my lack of balance and co-ordination problems with my legs and feet. Sometimes when I put my foot down my brain thinks it is secure on the step but in reality it has not made the step, I have had some falls because of this... falling is not reserved for stairs I have problems walking as well even when completely sober.” (Robert)
Some participants commented that negotiating such difficult environments "means I am looking down instead of being able to take in the world around me" (Robert). In this reflection, Robert alludes to missing out on experiences. This was further illustrated photographically and textually by Rose:

“As soon as you see uneven ground, slopes or steps you are immediately thinking I can’t do that...I have spent many a countryside or coastal holiday sat in the car whilst others have got out and enjoyed the view, beach, country walk etc. You get..."
to see what you can from the car window whilst traveling or from where you are parked. You put up and shut up even though inside you desire to be able to scramble down that rough track and enjoy life like everyone else.”

Visiting other people’s homes, shops, leisure facilities and even hospitals were also difficult due to issues with accessibility. Even where “accessible” environments were available, they were not always a solution:

“On the way I checked at the hospital for disabled parking places just before 9am - and every single one was occupied.” (Lynn)

“Bad things...long and difficult route to "accessible steps" (for those who can’t cope with a vertical metal ladder), competition with mothers and kiddies for the larger changing cubicles when there is nowhere to sit while waiting." (Lynn)

Lack of accessible accommodation was a problem for some participants whose basic needs were often not met. Photographing a shower stool she used at home, Kate commented:

“If I go away, I often go without a shower for one night as hotel bathrooms aren’t appropriately equipped and I don’t have the strength to get up out of a bath.”

(Kate)
For Lynn, difficulties going away were compounded by the challenge of travelling to get there: “Difficulty travelling really is a menace.” This challenge was further exacerbated by a lack of understanding and support for accessible travel:

“I bought my train tickets a few weeks ago- should have got them a week earlier (and £10 cheaper) but was misled by the ticket office man who assured me my power chair was too heavy for the ramp up to the train. Took me a number of phone calls to discover the actual weight limit of chair plus person is 300kg. When i returned and told him, he didn't believe me, but phoned a colleague in the station who agreed i was right. But he was correct about East Coast Main Line trains not taking wheelchairs in First Class (I had hoped for a bit of added comfort).” (Lynn)

This unaccommodating attitude was also depicted by Robert. Though he acknowledged that “the traveling public in general are very kind”, this was not always the case for transport staff:

“Once on the bus I face another problem of getting to my seat before the driver pulls away, using sticks means it is difficult to use the bus hand rails, even though I do ask the driver if he minds waiting till I have sat down most do but one or two do not. Then there is exiting the bus I always remain seated till the bus stops, and
even though there are plenty of signs around the bus telling you that is what you should do some drivers if they don’t see anyone standing to alight the bus they carry on."

Across a number of journal entries, Lynn described an on-going battle to fix the ratchet device which enabled her to transport her electric wheelchair, and consequently, access the outside world. Though many solutions were offered, paradoxically they were not suitable for someone with CMT. This left her feeling enormously frustrated and deflated:

“It is a nightmare and I feel as though I have gone down a snake (in life's game of snakes and ladders) - the problem was unreliable knots; the solution is virtually unusable - at least with my weak hands.”

In addition to navigating restrictive environments outside the home, participants also had to contend with similar challenges inside the home. Like many other participants, Robert characterised his every-day routine as permeated by frustration with himself and others:

“I am frustrated from the moment I get up; buttons, zips, socks, laces; it takes an eternity to get ready and is exhausting, sometimes I feel like exploding like the emoticon “Anger”.”
“Putting on anti-deodorant [sic] with their impossible tops that you have to push down to spray, and with the lack of grip, movement and dexterity of my hands and fingers it is an almost impossible task. I wish some of these manufacturers would consult less abled people when they design these products.”

Participants also discussed frustration and danger linked to other every-day activities such as cooking and carrying things:
“One of the negative impacts on your quality of life with CMT is kitchen frustration. All the jars, packets and pull tabs that you fight to get into. The different gadgets you buy and try to no avail cause your wrist and hand strength is rubbish... The pans you have to use two hands to lift because they are just too heavy and the risks of cuts and burns when your hands just give up and you drop it anyway... Not only is poor hand and wrist strength frustrating it is also an expensive fault in your design.” (Rose)

3.2.3 Main theme: Maintaining independence: Adaption, adjustment and support

This theme relates to the importance of maintaining independence and the different ways participants achieved this. The ability to maintain independence was described as fundamentally important to HRQoL. In order to preserve independence, participants had to make continual physical and psychological adjustments to maintain equilibrium as their condition progressed. Whilst adaptions and adjustments invariably enhanced HRQoL, they sometimes caused additional problems which were detrimental to HRQoL. Additional support to maintain independence came in many forms, including emotional and instrumental support from family and friends, some of whom also had CMT. Further instrumental support came from disability benefits and access schemes. Collectively, this support positively influenced HRQoL.
The fundamental importance of maintaining independence as far as possible was explicitly or implicitly evident in all participants’ journals. Lynn uploaded the below photograph and commented: “The power chair, vehicle and lifting mechanism are the result of a two year quest for mobility”. The bolded time period emphasises the lengthy battle she had faced to maintain her independence.

The photograph alludes to both how challenging and important this battle had been. This notion is reflected in the following account:

“The mud and bits of grass on the wheel are a souvenir of a very happy day out. I went recently with a friend to an annual potters' fair ("Potfest") held in marquees in the grounds of Scone Palace in intermittent torrential rain... I went last year with other friends who had borrowed a manual chair and took turns to push me,
which I appreciated enormously; but the wonderfulness of being independently mobile eludes description.” (Lynn)

For some participants, the act of taking part in the study itself was an assertion of independence:

“I am sorry that some pictures may be a bit fuzzy but I shake when I take them, and this journal is one thing I am doing myself.” (Robert)

The decision to under-go surgical adjustment was perceived as ultimately benefiting HRQoL and independence in the long-term by “reconstruct(ing) my feet into more usable things” (Jill). In the short-term, however, it adversely affected both. Rose uploaded a photograph of her post-surgery foot. The extent of the scarring reflects the scale of the operation and the associated loss of independence:
Rose’s reflective commentary provides further illustration of the extent of this lost independence and its impact:

“I have spent a total of 14 weeks in bed getting out to use a rotunda onto commode and needing 24 hour full personal care. A further 14 weeks learning to walk again with crutches and an aircast boot. 5 months on from my 2nd surgery I am still using crutches or a wheelchair when out and about depending on the distances involved. My life feels like it has stood still over the past year so this has definitely had a negative impact on the quality of life.”

Unsurprisingly, for some participants the loss of independence and risk of complications from surgery were too great a compromise to make. This decision reflects the importance of independence and autonomy for HRQoL:
“I also have a shoulder that I was told needs surgery. But I will put up with that because I don’t believe I can manage with no legs and only one arm after the surgery during rehabilitation and no one can guarantee that I will get the use of my arm back after surgery. I feel it is better to work with what I have than the unknown. The surgeon won’t have to live with the results but I will.” (Sarah)

In addition to surgical adjustment, several participants discussed the adapted self-management activities that helped them to maintain mobility and independence. These mainly involved physical activity:

“I really enjoy swimming .... I’m not very good at it and am very slow but try and add a length each week.” (Kate)

“Tuesday and Thursday are Water Exercises Days. XX Leisure Pool runs classes for people with medical conditions - some will get better, some are attempting to
fend off deterioration.-9.00 GP Referral Class, and 9.30 Maintenance Class. I attended the first for a year or so, before being "promoted" to the second” (Lynn)

Most participants also made proactive and pragmatic adjustments to their daily lives to accommodate CMT. These included creative solutions to every-day challenges and threats to independence:

“The picture is of my own personal solution to the problem of carrying the tea things from kitchen to garden and back. My tea "tray" has nice high walls enabling me to pile things up and not worry about spills - and a convenient handle over the top so that I can carry it in one hand, leaving the other free for a crutch (can manage with just one on the paved areas of my garden). Makes me feel rather smug.” (Lynn)
Over time, adjustments often included a change to employment. For example, John photographed his front room to represent the increased time he now spends at home:

“This year I had the opportunity of reducing my hours and I now work Wednesday, Thursday and Friday. I feel this has had a massive impact on my quality of life as I am able to spend my days off doing the things that used to swallow up my weekends and I have become less stressed about work problems.”

For many participants, the first step in making such pragmatic adjustments to activities and employment was accepting the need to do so:

“Do what I can during the day and try not to totally exhaust myself. I have learned to say “no” to new projects.” (Sarah).
Such acceptance was frequently challenging, however. For example, reflecting on her former employment in a Hospital, Jill wrote: “I have always worked and not in a job which I would recommend to "Me" Nursing, I spent most of my time with fatigue but overcome the problem by pretending it wasn't there.”

Where purpose built adaptions (e.g. orthotics and mobility aids) were required by participants, they often helped to enhance independence and HRQoL. Paul photographed two pairs of orthotics, thus indicating his reliance on them:

![Orthotics](image)

Paul’s description of these orthotics as “my best mates” mirrors the strength of feeling implied by another participant’s reflection: “As soon as I put these new orthotics on it was like walking with new feet and legs.” (Robert)
For some participants, however, accepting and adjusting to orthotics was challenging and the perceived benefits to HRQoL did not always outweigh the costs. Photographing her orthotics, Millie reflected that despite the fact they were new and much improved, she still experienced difficulties adjusting to them:

“However, they still feel weird, I still have to practice walking and I feel as though I’m having to constantly think HOW to walk and at 27 you don’t really want to be learning or retraining yourself how to walk.” (Millie)

Although they increased HRQoL, mobility aids also came with compromises. Lynn, for example, commented that needing space in a car for her electric wheelchair “reduced my choice virtually to a Ford Galaxy. This is a much larger car than I wanted,
but I wanted my freedom.” Adjusting to mobility aids was also associated with significant financial burden, particularly where Government support was lacking:

“We have the expense of having to buy an EnterVan with an automatic ramp if I want to drive my scooter into the van easily (cost $74,000)” (Sarah).

Most participants photographed and described their car as fundamental to independent mobilisation. Reflecting on the below photograph, Kate commented:

“After one of my operations I wasn't able to drive for over 6 months... I had to rely on others. So I really appreciate my car and my independence. The first time I drove again on my own I actually cried with joy!”
The majority of participants conceptualised their independence as contingent upon the support they received. This took many forms, ranging from instrumental to emotional support.

For some participants, disability benefits were vital for accessing a car and the associated positive impact on HRQoL:

“One of the things that has made an absolute improvement on the quality of my life with CMT is the benefit Disability Living Allowance. I have received higher rate mobility allowance since I was 25 years old, I am now 48. This benefit has allowed me to be totally independent when I need to go out.” (Rose)

Access to a disabled badge was also described as having a “positive outcome on the quality of my life” (Rose). This was due to having more space to get in and out of cars and reducing walking distances, thus increasing independence:
“I was able to park near the shops and then the Vegan cafe, thanks to my Blue Badge. This helped immensely as I tire easily.” (Kate)

Most participants illustrated the way in which their family and friends, particularly their partner, supported them to maintain independence and positively influenced their HRQoL. This was both instrumentally and emotionally:

“My husband is really understanding about CMT. He helps me a lot and is used to me coming in with a jar for him to open, or sitting back down several times when trying to get up from a chair and wobbling around. He carries things for me and holds my hand .... not to be romantic but to pull me along or keep me upright! He is also emotionally supportive during the (quite rare) occasions when I get upset.” (Kate)
Millie photographed herself holding hands with her partner. This is a powerful visualisation of unity and support:

“The negative aspects which can affect quality of life are negated by the fact I have somebody there who I know loves me and is willing to push me around in a wheelchair if (heaven forbid) it ever got to that stage and he makes my quality of life much more positive.”

The fundamental role of partner support was poignantly illustrated by recently widowed Lynn, who reflected on a future without her husband: “This is when living
alone is a huge problem. Think I will have to depend on carers popping in, and will really have to try to crack the problem of buying supplies online.”

Some participants discussed the emotional, instrumental and informational support they shared and received with other family members who also had CMT:

“I have to say that I have two daughters who are really helpful. They live nearby and take me to the beach or cinema to help me get there. I also have 5 sisters and my Mum. We all help each other in different ways ... Mum needs more help now and whilst I can’t help physically I order things for her on the Internet or drive her to appointments. Having CMT we can share ideas and support each other. All my family were great support following my operations. They really improve my quality of life.” (Kate)

3.2.4 Main theme: Lack of control and future-orientated anxiety

Many participants described and illustrated a perceived lack of control over CMT and experienced anxiety. This anxiety related to coping with the challenging and progressive nature of CMT, as well as threats to the external assistance participants received to help them to manage the condition. The risk of passing on the condition to
children and grandchildren was a heavy burden faced by many participants, leading to further anxiety and difficult decisions.

Some participants described significant anxiety related to their perceived ability to control and cope with the consequences of CMT. For Rose, a lack of perceived control and ability to manage her unpredictable pain led to the experience of panic attacks:

“I went through a period of panic attacks and anxiety due to the stress of worrying about pain striking and my ability to cope with it if out and about. The anxiety and panic reached such a level that it permeated other parts of my life so that not only would I get in a panic if I had to stand in a queue, worrying about pain and feeling like I'm going to pass out to then starting to get panicky if I had to queue in a traffic jam. It was so bad one weekend that I thought I was having a heart attack and was going to die…” (Rose)

Many participants discussed the anxiety they felt in visiting new or unfamiliar places. This was principally in relation to whether they would be able to cope, and a perceived lack of control in these situations. Reflecting on a photograph of a swimming pool John commented:
“Going to places like this with the wheelchair gets me very anxious as I don't know what facilities they will have or how to access them.”

At times, the anxiety associated with new and unfamiliar surroundings actively prevented participants from engaging in activities:

“With CMT you worry a lot and build hurdles to prevent you from doing things. For example you are invited somewhere new, say for dinner. You instantly worry, will you be able to park close by, will there be stairs or steps, will there be a banister to hold onto? Will the seats be low, will you be able to get up? Will the toilets be upstairs, will there be a disabled toilet? As such quite often you talk yourself out of doing things so as not to have to worry about them because if you start to worry, you will start to stress then you won't be able to get up from the seat, you will stumble into everyone and generally end up looking like the worst sober drunk at the party. Stress and worry have a real negative effect on CMT and as such you find you are happiest when at home and have to push yourself to get out and do something different.” (Rose)

This extract illustrates the anxiety Rose experienced about coping with the challenges imposed by her condition in an unfamiliar setting. As well as worrying about
her abilities, there is also the suggestion that Rose is worried about what others might think. The perceived lack of control over her condition in unfamiliar situations generated significant anxiety and, ultimately, avoidance.

Anxiety about coping with CMT itself was sometimes exacerbated by threats to the benefits and external assistance participants relied upon to manage the impact of their condition:

“Very much the worry at present is that when my DLA is replaced with Personal independence Payment will the person assessing me understand my condition and will I be able to keep my independence. We cannot afford to run a vehicle for me without this benefit... The stress and worry about the future and any changes in benefit will have a severe impact on my quality of life making me housebound during the working week and killing my social life which keeps me from having time to dwell on things.” (Rose)

The reality of this anxiety was illustrated by John, who received his benefit assessment letter during the course of his participation in the study:
“I had hoped that I wouldn’t have to do the face-to-face assessment until March or April, but my appointment letter arrived yesterday. It put me in a bad mood all evening and impacted on the way I interact with my wife and children. I know there is nothing I can do about this change, but it doesn’t seem to matter how much I tell myself this, it remains a big worry. It is booked for the 14th February, so at least it will be over with fairly quickly, but at the moment all I can think about is the impact the loss of this benefit will have on my daily life.” (John)

This photograph and associated extract both highlight the anxiety provoking nature of the current benefit reforms in the UK. Not only did the letter appear unexpectedly, finding out that a potentially life altering assessment will take place simply “with a Health Professional” (as detailed in the photograph) introduces a further element of uncertainty. The sense of lack of control over this process is powerfully illustrated in John’s words: “I know there is nothing I can do”.
As well as the worry of losing vital support in the future, participants were acutely aware of the progressive nature of CMT. For example, Jill reflected that “my hands have weakened much more over the past couple of years...”. Another participant commented on the progression he noticed after a trip to his local swimming pool one day: “This visit to the pool also served to remind me how much weaker I am than a year ago.” (John)

For most participants, CMT progression and the associated lack of control led to anxiety about what life would be like in the future. When first diagnosed, Paul worried he would lose the ability to continue with an activity he loved:

“I’ve always enjoyed cycling especially off road terrain riding (I hate tarmac) and at first when diagnosed with CMT and knowing it would lead to muscle
deterioration in my calves I was worried. Would I still be able to continue with one of the greatest joys in my life or not?” (Paul)

Recently diagnosed Jessica worried she may not be able to maintain her voluntary role with the coastguard as her condition progressed:

“It feels good to train to do this means I am doing something so positive… but also comes with a sadness as I don't know how long I will be able to do this for. Will my cmt mean I have to give it up…” (Jessica)

As CMT progressed, participants also worried about coexisting conditions and how they would cope when even basic activities became difficult:
“The hip/arthritis moan makes me think ahead to when I can't cope physically and would need to consider having a live-in carer. But how on earth would I begin to find someone suitable? Also I would need to clear a room to accommodate them...

There are an awful lot of things I am going to have to sort out, but it is so hard, physically and emotionally.” (Lynn)

The familial nature of CMT meant that some participants witnessed more advanced stages of the condition in their relatives. This compounded anxiety about the impact of progression, both for the participants themselves and their loved ones.

Reflecting on a photo of her Dad, Jessica wrote:

“Dad: Don't want to lose you to this fucking condition ..... Doing what you love sailing and helping you still to do it is easy, watching the possibility that you won't breaks me.”

The familial nature of CMT led to further uncertainty and anxiety related to passing on the condition to offspring, and most participants discussed inheritance as a substantial burden. Paul’s diagnosis of CMT was relatively recent. His final journal entry concluded with a photo of his Grandchildren and the following poignant reflection:
“Finally I just hope and pray that I have not passed on CMT to them or my daughters. That would devastate me completely.” (Paul)

Anxiety at the prospect of having unknowingly passed on the condition implicit in this statement was mirrored by the anxiety some participants felt in making reproductive decisions with a known diagnosis of CMT. Whilst some participants “went to a geneticist about the impact of this on having children” (Millie), other participants decided not to have a family due to the risk of inheritance and a perceived lack of control over this. Jessica photographed a pram to reflect a future she could not imagine for herself following her diagnosis of CMT. Her reflection poignantly illustrates an emotive response to the threat of CMT inheritance and a perceived lack of control over her choice to have children or not:

“Baby in the house...
And it's not mine and because of this stupid condition it will never be part of future. No words.”

3.2.5 Main theme: CMT awareness

Many participants identified a general lack of awareness about CMT, including amongst health professionals. This led to delayed diagnosis and management, and also compounded the difficulties participants faced in disclosing their condition to others. Thus, lack of awareness negatively influenced HRQoL. In contrast, greater awareness and professional support was valued by participants and associated with improved HRQoL.

Many participants commented on the lack of knowledge about CMT amongst medical professionals. This was detrimental to HRQoL and perpetuated a lack of appropriate healthcare support:

“Medical staff are another problem, when you are asked if you have a long term medical condition and my reply is CMT, I like to observe the look on the face to see the blank expression.” (Robert)

For some participants, this lack of awareness led to lengthy delays in diagnosis and appropriate management:
“I was diagnosed in my late 20's this seems rather late when you think I had always had foot problems along with falling and balance problems from a very young age.” (Jill)

“Now I know about the condition I have had this since childhood altho it took till I was over 50 to get a diagnosis.” (Helen)

Indeed, some participants felt compelled to raise awareness and understanding of CMT within the medical profession to improve diagnosis and management. Patient organisations such as CMT UK were felt to be particularly helpful:

“I am a member of CMT.org and have found the literature they have produced useful to try and educate the various medical people I have encountered during my life.” (Robert)

“I am now an authority and have had my GP and all the surgeons I worked with read up and contact CMT UK for information just in the hope if it effects my grandson or anyone else's a little more will be know and diagnosis will be more speedy.” (Helen)
Where healthcare support did exist, it was highly valued by participants:

“I feel positive to have the support of a podiatrist.” (Kate)

“I am very lucky to have a relatively young doctor who is very clued up on my condition and a good orthotics and podiatrist team who look after me. This has
not always been the case and over the years I feel that I have had a fight with the medical profession to get people to listen.” (Robert)

A general lack of awareness amongst most healthcare staff was perceived to inhibit understanding about CMT in the general population, making it difficult to tell others about the condition:

“As a nurse with extensive orthopaedic training I was in the dark when given the diagnosis so what hope have non-medical patients?” (Helen).

“Very few doctors have ever heard of Charcot Marie tooth so try explaining it to a typical person and expecting them to understand.” (Millie)

Disclosing the condition to others was discussed by many participants as inherently challenging. In part, this was due to the fact the condition is unknown and difficult to understand:

“I used to try and explain my condition which is very difficult as it is the most common unknown condition of all time (my opinion). Now I just say I have a
Neurological Muscular disease and smile sweetly and I find that normally suffices.” (Robert)

“To begin with I played it down, then when it became more serious, I tried to explain it in more detail, however it was hard. The specifics sound horrendous at times and trying to explain something that you don't 100% understand yourself is hard too.” (Millie)

Early on in the condition, some participants chose not to disclose their diagnosis to others:

“Only two on the team know of my diagnosis will I be able to keep this secret will anybody notice the changes in my ability.” (Jessica)

Difficulties in communicating a CMT diagnosis were further aggravated by the fact that, for many participants, the condition was relatively invisible, most of the time:

“One of the main problems is about letting people know you have a problem with mobility and balance. If I'm wearing my orthotics and dressed in long trousers
then most people would have no idea of my problem. They might notice a slight stiffness in my walking style but in most situations it is not obvious.” (Paul)

In addition to making CMT difficult to explain, misconceptions about invisible illness were a source of frustration for participants:

“The only thing I really get annoyed with is when people can’t see a visible problem they assume there is nothing wrong with you.” (Robert)

For some participants, mobility aids were a helpful way to visually represent the condition and alert others:
“One technique I use in crowds, like airports, concerts etc is to take a walking stick. See first photo. It’s the most obvious way to indicate I have a problem. Most people recognise this and give me space and time.” (Paul)

This was not an acceptable solution for everybody, however, with one participant commenting that they “disliked immensely” (Kate) their experience of needing to use a walking stick. For many participants, the need for understanding and assistance was juxtaposed with the need to blend in. In other words, a tension existed in balancing visibility and invisibility.

3.2.6 Main theme: Threats to identity and self-esteem

Loss (and anticipated loss) of valued roles and associated identities generally had a negative impact on participants’ self-concept and self-worth. For some participants, embarrassment detrimentally affected self-esteem. Women in particular worried about their appearance and felt frustrated by their limited and unfeminine choice of footwear and clothing. Thus, the experience of CMT threatened some participants’ feminine identity. Threats to identity and self-esteem negatively influenced HRQoL.

Like a number of other participants who had lived with CMT for some time, Rose discussed giving up work within her journal. Despite her photo representing the
substantial financial burden imposed by loss of employment, her reflection also highlighted the detrimental impact of finishing work on her identity and self-esteem:

“My sense of who I am also took a hit, going from college lecturer to homemaker. My world suddenly became smaller as I wasn’t out seeing people on a regular basis and I felt quite useless and annoyed with myself because I just could not do it anymore.” (Rose)

Anticipated loss of occupational identity was also evident within some participants’ accounts. Part of Jessica’s sadness at the possibility of not being able to maintain her voluntary role with the coastguard was the anticipated loss of this important aspect of her sense of self. The anticipated loss of occupational identity was further illustrated in another reflection about her business and business partner:
“How do I tell you that I might need to give this up to life my life before it gets me.

No words to describe the sorrow.” (Jessica)

Other participants reflected on threats to their artistic and creative identities:

“I don’t listen to so much music nowadays. Maybe because it reminds me that I had to give up singing in the XX Choral Society in 2012 because of a voice problem - possibly caused by CMT. But I’d never get up on to the Concert Hall stage for concerts now anyway. Last time I took part, I couldn’t stand safely and had to sit on a high stool. I was very sorry indeed to give up.” (Lynn)

“I am a writer. I can no longer hold a pen or pencil for any length of time or type. I have gone from two finger typing to weaving small pencils in and out of my fingers to stiffen them to hit the keys, to typing with the knuckles of my little fingers and now to using speech recognition software on my Mac which is a godsend.” (Sarah)
Identities within relationships were also threatened at times, as participants felt unable to conform to normative roles. Reflecting on his trip to the swimming pool, John commented:

“My wife helped a lot as usual, but I feel sorry for her as sometimes it must seem as if she is caring for 3 children rather than just 2.”

This perceived inability to conform to normative roles extended to other relationships. For example, two participants discussed the impact of CMT on their identity as a Grandparent. Reflecting on a photograph of his Grandchildren, Paul commented:

“I can't run around with them, play active games with them, help them on adventure playground activities etc, etc. I'm even restricted in not being able to go swimming with them... The elder ones do understand about Grandpops poorly feet and make allowances for me but it still hurts me when I think of the things I can't do for them or with them.”

For some participants, dissociation from a “disabled” identity was important for self-esteem. Reflecting on her raised toilet seat, Rose commented:
“Also I feel like a "normal" person because I don't have to have disabled toilet aids on view for all to see...”

Later on in this journal entry, Rose described embarrassment related to using other people’s toilets:

“Using other people's toilets or public ones when disabled aren't available impacts negatively on my quality of life as it is embarrassing when after several attempts to get up you have to ask for help or you hurt your back twisting to hold onto the cistern to get up.” (Rose)

For some participants, historical experiences of embarrassment influenced responses in the present day:
“Even though the tutor stresses it is "just for fun", somehow when we all have to walk to the other side of the pool and back (water's up to my armpits), and I am last by a very long way, it reminds me uncomfortably of PE lessons at school (which I didn't just detest: I dreaded them.” (Lynn)

Appearance concerns were particularly salient for women and the majority of female participants discussed the complex way in which CMT interacted with their feminine identity. Though some commented that they “largely ignored” (Jill) the looks they received from others, most were self-conscious about their appearance:
“I wear trousers at work and in public but at home or on the beach I wear shorts and feel self conscious about my legs.” (Kate)

“No plans to see anyone, so wearing cool summery mid-calf trousers - which show my AFOs and boots (more clunky than cool or summery)... She knows about my legwear, so I didn’t mind her seeing it.” (Lynn)

Furthermore, restrictive footwear and clothing often prevented women from feeling feminine and had a detrimental impact on their self-esteem:

“This is one for all the ladies with CMT. Oh what a joy it would be to wear pretty, dainty shoes with a kitten heel. No, we have to wear clumpy, ugly wide fitting sensible shoes over our mottled skin, deformed and ugly feet (even after corrective surgery). Part of a good quality of life is being able to dress up for any occasion, as
a woman to feel feminine and sexy. With CMT you generally have very little option of what will fit your feet, it will be clumpy, it will be a wider fit and it will not have a heel. For over 7 years I had the choice of one pair of hospital, custom made shoes to fit over splints to wear for every occasion, pegging washing out on muddy grass to a posh evening out. Irrespective of what I chose to wear the shoes were only in one colour, in my case some years purple, some years navy. One pair per year.” (Rose)
“...she's good really good but she can't stop my nails from being disgusting oh to wear flip flops and nail polish it's a simple think in many girls life's just not mine !

Another summer in shoes that hold my orthotics and hide my toes.” (Jessica)

3.2.7 Main theme: Resilience and positive growth

All participants expressed considerable resilience and positive growth within their journals. As part of this, participants expressed gratitude for the things they were able to do, in spite of, and because of, CMT. Maintaining valued roles and activities and connection with others was perceived as fundamentally important. Together these positive psychological factors helped participants to re-frame their experience of CMT and enhance perceived HRQoL.

Many participants discussed CMT as being “very much what you do with it” (Jill). A determination not to be constrained by CMT was evident across participants’ narratives:

“Nevertheless I don't allow this to stop me trying I have always just got on with it... I think the problems I have do affect my quality of life negatively to a certain
extent, but if I think of life’s challenges as positive to my outlook on life then I won’t be disappointed.” (Jill)

“It is what it is. My philosophy is to do the very best I can with what I have for as long as I am able and to never stop learning or giving.” (Sarah)

For some participants, family members with CMT were an inspiration and model for such resilience:

“...you have taught me how to drive through this hard without doubt and how you face the challenge of your life. But yet you still pitch up and try to keep going no matter how hard.” (Jessica)

This determination led to the identification of positive coping strategies which helped participants to develop a sense of mastery over current and future challenges:

“I can say that every day presents a new challenge but with that also comes a new coping strategy.” (Robert)
“For the future as and when it gets more difficult to sustain this pattern of rides I've my eye on an electric bike!” (Paul)

The importance of not being defined by CMT was evident across participants. Millie, a 27 year old teacher reflected that “CMT is just a part of me, it isn't all of me.” This sentiment was echoed by Sarah, a painter and writer:

“However, I am not my CMT. I have a brain that is able to grasp, absorb, calculate and retain. I am curious and creative and that has nothing to do with CMT however it helps me cope with the syndrome and has open the world to me...I have senses that allow me to enjoy a great many experiences that the world has to offer exclusive of mobility and detailed dexterity.” (Sarah)

Determination to maintain competence in valued activities and roles was often matched with gratitude and appreciation. John photographed the handcycle he now uses instead of a standard bike. It represents his ability to continue cycling and the freedom he is grateful to experience:
“I worked out I was able to buy a very basic Handcycle using the cycle to work scheme and my handcycling hobby started. I have since upgraded to the model in the picture and I spend as much of my free time on this as I can...The sense of freedom I get whilst cycling is great and even though I am very slow, I still enjoy it immensely.” (John)

Two participants identified themselves as artists. Whilst Sarah could no longer paint detailed botanicals, she expressed positive growth, finding enjoyment and appreciation of new found abilities in abstract painting:
“I am a painter. I studied art for three years before I studied psychology. I used to paint very detailed botanicals but I can no longer hold and control small brushes. I now enjoy the use of bold color on large canvases and paint abstracts with large brushes and tools that I can hold with two hands. They are much more difficult than botanicals.”

In contrast, Lynn could still paint extremely detailed botanicals. She made social comparisons and considered herself fortunate compared to others:

“Made me appreciate even more how lucky I am to be able to do extremely precise drawing...in spite of my CMT.”

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Signature covered to protect anonymity. All images are subject to Copyright.
Some other participants also made self-enhancing social comparisons to identify positives in their own experiences:

“I do sometimes wish I could still walk the cliff paths rather than sitting with a book, but realise I’m fortunate to still be able to do as much as I can.” (Kate)

“While I cannot say I would wish to have CMT if given the choice, I am fully aware that I could have been born with something far worse.” (Sarah)
Other participants described benefit finding, reflecting on the achievements and experiences that specifically related to having CMT:

“On days when my disability is getting me down, I try to remind myself of the doors it has opened for me and the things that I would never have had the chance to do without it.” (John)

“CMT and my curiosity and talent has given me status that I might never have attained had I not had the disease...Simply knowing that my work regarding disability issues and people with CMT is valued by my peers has kept me going many times when my enthusiasm and energy flag.” (Sarah)

The importance of relatedness was also evident across participants’ accounts. Indeed, many participants photographed the people they valued. Many participants reflected the notion that “the person makes this life so much better!! (Jessica). For example, in the journal entry “pros and cons” Sarah reflected: “I have a soul that affords me the empathy I need to connect with my peers. I love and am loved and consider that huge.” In an earlier journal entry about losing the use of her hands, Sarah discussed what she was still able to do: “I can still hold my husband’s hand, touch his
face and feel his warmth.” Thus, physical and emotional connection with others was important for HRQoL.

Lynn regularly expressed gratitude for the friends and supportive people in her life: “I am very lucky with my friends.” The photograph and extract below illustrates the concept of relatedness and its positive influence on HRQoL:

“We talk about all sorts of things and watch birds through the kitchen windows, and sometimes don't disperse until well after 2. I think it is the highlight of my week. Today there were just 6 of us, sitting under an umbrella in the warm sunny garden. We are like old friends now.” (Lynn)
Maintaining independence: adaption, adjustment and support

Lack of control and future-orientated anxiety

Threats to identity and self-esteem

Resilience and Positive Growth

CMT Awareness

The challenge of the every-day

Physical challenges of CMT

Navigating restrictive environments

Positive and negative impact on HRQoL

Positive impact on HRQoL

Negative impact on HRQoL

3.2.8 Figure 1: Thematic map
4. Discussion

The aim of this study was to explore how adults living with CMT understand HRQoL and the factors which influence this. It was hoped that the photovoice method utilised in the present study would enable the exploration of the dynamics of HRQoL, as it is experienced by adults with CMT. The findings from the present study will be discussed with respect to the aim and objectives of this thesis, previous research in this field and relevant theory. Strengths and limitations of the current study will be presented and implications for future research and clinical practice will be highlighted.

A number of common themes emerged across participants’ photographs and textual descriptions. As anticipated, participants’ experiences revealed important nuances in every-day experiences of HRQoL. Thematic analysis of the visual and textual data identified 6 main themes related to HRQoL: “The challenge of the every-day”, “Maintaining independence: adaption, adjustment and support”, “Lack of control and future-orientated anxiety”, “Threats to identity and self-esteem”, “CMT Awareness” and “Resilience and positive growth”. All participants discussed living with CMT as challenging and frustrating. This was often due to an interaction between the various debilitating physical manifestations of CMT and the physical and social environment. Maintaining a level of independence was important to participants who had to make continual adjustments, use adaptions and seek support to re-establish a state of
equilibrium, as their condition progressed. The progressive, uncertain nature of this hereditary condition led many participants to perceive a lack of control over CMT and to feel anxious about the future. A lack of awareness about CMT in both the medical profession and the general public compounded the difficulties participants faced in disclosing a largely invisible condition and seeking support. Furthermore, experiences of difference in childhood and adulthood adversely influenced self-esteem and self-concept for some participants. Sense of identity was generally threatened by loss of valued and normative roles. Additionally, limited accessible footwear, restricted clothing and visual difference hindered women’s sense of femininity. In spite of the many challenges participants faced, all of them demonstrated considerable resilience and positive growth in the face of adversity. This enabled participants to re-frame their values and expectations and to find meaning in their experiences. Engaging in valued activity and connecting with others were frequently conceptualised as key goals for HRQoL.

4.1 Understandings of HRQoL

Together the identified themes highlight that for adults with CMT, HRQoL is a complex and multi-faceted concept (Bakas et al., 2012). Participants were not asked to define HRQoL, instead their understandings were deemed to be implicit within the experiences they chose to illustrate. All participants photographed and wrote about the physical aspects of their condition and the way in which they made life difficult.
Importantly however, not only was there variation within the objective physical manifestations of CMT, there was also variation in their perceived impact, both within and between individuals. For example, the numerous physical consequences of CMT appeared to be differentially problematic across the two-week period. In some entries, participants described experiencing particularly high levels of pain or fatigue that day or feeling particularly frustrated by their lack of strength (within individual variation). Furthermore, although difficulties with independent mobilisation were frequently discussed as detrimentally impacting HRQoL, loss of hand functioning was perceived as more devastating by one participant who identified as a writer and painter (between individual variation). Thus, HRQoL was highly individualised and inextricably linked to participants’ values. Moreover, HRQoL was more than the physical manifestations of CMT and their respective impacts on activities of daily living and social and occupational functioning. Participants illustrated a range of psychological consequences of living with CMT such as stress and anxiety, low self-esteem and an impaired sense of femininity. Additionally, participants discussed the process of accepting their limitations and adjusting to CMT as key components of HRQoL. Although highly individualised, attaining HRQoL generally emerged as the ability to accept and adapt to the physical aspects of CMT, with appropriate support, in order to engage in valued activities, hobbies and occupational pursuits, to achieve a sense of coherence and positive growth and to sustain relationships with important others. This conceptualisation is consistent with the Disability Centrality model which suggests that the impact of a health condition on quality of life is dependent upon the extent to
which it reduces satisfaction in central domains, either by inhibiting perceived control or by adversely affecting the ability to engage in valued roles, activities and relationships (Bishop, 2005).

4.2 Factors which influence HRQoL

4.2.1 Negative influences. As identified in the thematic map (see Figure 1) HRQoL was subject to a number of different negative influences. The theme “The challenge of the every-day” captured the numerous every-day challenges faced by participants which were perceived to detrimentally influence HRQoL. Despite variation in disease type and progression, all participants highlighted a range of “physical challenges of CMT”, including reduced mobility, strength, balance, dexterity, sensation, pain and fatigue, which made every-day activities difficult. Previous research has often focused on the physical aspects of HRQoL in adults with CMT and has highlighted a range of physical predictors of impaired HRQoL (Roberts-Claude et al., 2016; Johnson et al., 2014, 2013; Boentert et al., 2010; Redmond, Burns and Ouvrier, 2008; Padua et al., 2008, 2006). Johnson et al.’s (2014) study, for example, indicated that foot and ankle weakness and limited mobility were perceived as having the most adverse impact on HRQoL across a broad range of physical symptoms. The current study did not seek to quantify the relative importance of physical aspects of CMT, but rather to explore these experiences in depth. Whilst foot and ankle weakness and limited mobility were found to negatively affect perceived HRQoL in the current study, loss of hand/ wrist
functioning, poor balance, pain and fatigue were also perceived as extremely challenging and stressful. Thus, the current study adds to the existing literature highlighting the adverse impact on HRQoL of a broad range of physical manifestations of CMT. The study also highlights the complex, differential impact of the physical manifestations of CMT. This relates to both individual differences in objective disease indicators and, importantly, individual differences in perceived impact on HRQoL. This may explain why discrepancies have been found in previous quantitative studies regarding the relative importance of physical predictors of HRQoL (Roberts-Clarke et al., 2016; Johnson et al., 2014, 2013; Boentert et al., 2010; Redmond, Burns and Ouvrier, 2008; Padua et al., 2008, 2006).

Importantly, participants also documented difficulties in “navigating restrictive environments”. These included, for example, uneven terrains, stairs, public transport and leisure activities. Participants felt frustrated at missing out on experiences and by the challenges associated with activities of daily living. Sometimes these were perpetuated by a lack of understanding or thought from others. The “challenge of the every-day” was thus often a consequence of an interaction between the physical manifestations of CMT and restrictive physical and social environments. Whilst the physical manifestations of CMT have been the subject of much research (Roberts-Clarke et al., 20016; Johnson et al., 20014, 2013; Boentert et al., 2010; Redmond, Burns and Ouvrier, 2008; Padua et al., 2008, 2006), interactions with the physical and social
environment have been largely overlooked. Limited qualitative research has previously
alluded to such an interaction in adults with CMT, but this study did not provide
supporting evidence to substantiate the claim, or explore this concept in depth
(Lafarge et al., 2014). Therefore, this is the first study to provide in-depth visual and
textual accounts of such interactions from participants’ every-day experiences. As
such, the current study extends previous knowledge and illustrates the broader
influences on HRQoL, which have been omitted from previous research.

The every-day challenges participants faced were perceived as inherently stressful.
Moreover, stress was perceived to exacerbate the physical manifestations of CMT.
Thus, stress was both an outcome and an influencer of HRQoL. Previous research
suggests that daily hassles, or every-day stressors, inversely predict adjustment in
chronic disease in young people and adults (von Weiss et al., 2002; Thompson et al.,
1992). Support to navigate the physical, social and environmental barriers to HRQoL is
therefore important to facilitate adjustment in adults with CMT.

Perceived difficulties in managing a progressive, hereditary condition, in
combination with threats to external support, led many participants to experience a
“lack of control and future-orientated anxiety”. Participants worried about how they
would cope in new and unfamiliar surroundings, as well as how they would cope as the
condition worsened and valued activities became difficult. Inheritance was perceived
as a heavy burden and was a source of anguish to participants with children and
grandchildren, as well as those who were deciding whether to start a family. Thus,
perceived lack of control and future orientated anxiety adversely influenced HRQoL.

Previous quantitative research has reported mixed findings on whether individuals with CMT experience higher levels of anxiety than healthy controls, and individuals with other types of neuromuscular disease (Cordeiro et al., 2014). In contrast, existing qualitative research has previously identified anxiety as problematic, specifically in relation to fear of progression and inheritance (Lafarge et al. 2014; Alberts, 2008; Arnold, McEntagart and Younger, 2005). The current study supports and extends existing qualitative research, suggesting that adults with CMT experience specific, daily anxieties related to the broad impact and management of the condition. These include the possibility of losing vital external support such as mobility assistance payments.

In the United Kingdom, the introduction of the Personal Independence Payment (PIP) has meant that millions of people in receipt of a disability benefit, formerly Disability Living Allowance, have been re-assessed or are currently waiting for re-assessment (DWP, 2017). The driver for this change was in part financial since a 20% reduction in claimants was set out as a government objective when the scheme was first launched (Henry, 2014). There is a lack of systematic evaluation of the impact of this benefit change (Machin, 2017); however, challenges negotiating the process have previously been reported for individuals with other conditions (Davies et al., 2017). The current study highlights the anxiety this process causes adults with CMT, who fear losing this entitlement and, consequently, their HRQoL. Whilst the rhetoric surrounding disability benefits is largely around access to employment, the importance of these benefits for maintaining HRQoL must not be disregarded.
Within the theme “threats to identity and self-esteem”, many participants discussed losses of functioning and abilities as negatively affecting their self-concepts. For some participants, these experiences led to low self-esteem which adversely affected HRQoL. Issues with self-esteem have previously been identified in some qualitative studies (e.g. Lafarge et al., 2014; Alberts, 2008; Arnold, McEntagart and Younger, 2005) and personal reflections (Crabtree, 1997) about CMT. The current study suggests that low self-esteem is not an inevitable consequence of CMT but can be problematic for some individuals. Despite some evidence to suggest that onset of illness in later life is more likely to affect self-esteem than illness which begins in childhood (Bishop, 2005), this distinction did not emerge between participants in the current study whose onset of CMT and age at diagnosis varied greatly. Moreover, some participants spoke of traumatic early experiences which stayed with them in adult life, such as being unable to keep up with others in Physical Education (PE). Future research could therefore usefully examine longitudinal predictors of self-esteem in adults with CMT.

Participants in the present study had to adapt to lost or altered identities as their condition progressed. Whilst loss of occupation resulted in obvious social and financial implications, importantly, this loss and the associated implications also affected participants’ sense of identity. The current study supports previous quantitative and
qualitative research highlighting the adverse impact of unemployment on HRQoL in adults with CMT (Johnson et al., 2014; Lafarge et al., 2014; Vinci et al., 2005). It also suggests, however, that this loss can impede an individual’s sense of identity, which independently affects HRQoL. Moreover, the current study highlights the importance of creative and sporting identities. Threats to these facets of identity also appear to detrimentally influence HRQoL. Multiple threats to identity been identified in qualitative studies exploring the lived experience of chronic illnesses (e.g. see Roing and Sanner, 2015 for a meta-ethnographic synthesis). However, the concept of identity has been largely ignored within existing CMT research which has thus far only recognised rejection of, or confusion surrounding a disabled identity (Lafarge et al., 2014; Alberts, 2008).

Threats to gendered identities also appeared to detrimentally influence HRQoL. Women in particular discussed appearance related threats to their feminine identity. Masculinity and femininity norms are thought to influence the lived experience of chronic illness. Whilst men are socialised to view their bodies as functional, women are socialised to identify their bodies with aesthetic evaluation and beauty. Thus, chronic illness may undermine men’s ability to conform to hegemonic masculinity and women’s ability to attain idealised standards of beauty and, thus, femininity (Clarke and Bennett, 2013). The current study suggests CMT-related differences to the feet, hands and legs, and associated restrictions to clothing and footwear, may adversely
influence women’s sense of femininity. Although body image concerns have been alluded to in some quantitative and qualitative CMT studies (Johnson et al., 2014, 2013; Alberts, 2008; Arnold, McEntagart and Younger, 2005), the link with feminine identity has not been identified. Concerns about appearance and difficulties wearing feminine clothes and shoes adversely affect HRQoL and may be important influencers for non-adherence to orthotic devices and/ or surgical intervention in women. Indeed, previous research has found appearance concerns to be an independent predictor of desire to undergo corrective surgery in women with arthritis (Vamos, 1990). The impact of CMT on femininity should therefore be recognised as an important determinant of HRQoL. Furthermore, future research should explore the impact of CMT on masculinity; this was not apparent in the current study, potentially due to the small number of male participants.

Collectively, “the challenge of the every-day”, “lack of control and future-orientated anxiety” and “threats to identity and self-esteem” were all associated with adverse effects on HRQoL. Inter-relationships between these themes were apparent, for example the every-day challenges and frustrations participants faced contributed to a sense of loss of control and anxiety about coping, both in the here and now and in the future. Furthermore, these every-day challenges and the associated loss of control threatened participants sense of identity and self-esteem.
4.2.2 Negative and positive influences. As anticipated, differences between and within individuals led to the identification of key themes which both negatively and positively influenced HRQoL. The theme “maintaining independence: adaption, adjustment and support” highlighted the importance of independence and the way in which participants had to adjust, physically and psychologically, to maintain their independence and partake in valued activities. As their CMT progressed, participants had to make constant adjustments to activities and use adaptions to maintain a level of independence and re-establish equilibrium. Support from social and community networks and Government-backed mobility schemes helped to facilitate this. Whilst adaptions and adjustments often enhanced independence and HRQoL, they were sometimes difficult to accept and/or led to compromises and further losses of independence. Consequently, HRQoL was sometimes adversely affected by adjustments and adaptions, at least in the short-term. Previous research has largely overlooked the importance of maintaining independence in adults with CMT. Limited qualitative research has highlighted loss of independence within the lived experience of CMT (Lafarge et al., 2014; Arnold, McEntagart and Younger, 2005). The concepts of “finding alternative ways” and adjustment have also been identified (Lafarge et al. 2014; Alberts, 2008 p. 292). Additionally, one qualitative study previously highlighted the practical and cosmetic concerns that AFOs present to individuals with CMT (Phillips, Radford and Wills, 2011). Whilst the importance of support has been identified previously, this has typically focused on immediate social support and has not included the broader sources of instrumental support (e.g. mobility assistance...
schemes) (Lafarge et al. 2014). The current study therefore extends previous knowledge, highlighting the value adults with CMT place on maintaining independence, the nuanced impact of making adjustments and using adaptions and the broad context of social support.

Despite many pros and cons to most adaptions and adjustments, the current study found that adults with CMT perceive that adapted exercise and physical activity positively influences their HRQoL. This was in spite of some evidence of unpleasant, shaming experiences in PE as a child. The links between physical activity and quality of life have previously been established in individuals with Muscular Dystrophy (McDonald, 2002). The influence of physical activity on HRQoL for people with CMT has however been thus far overlooked. This may therefore be an important direction for future research and clinical practice.

The theme, “Awareness of CMT” also had the potential to both positively and negatively influence HRQoL. Due to a lack of awareness about CMT in the health service and general population, participants frequently experienced difficulties in gaining a diagnosis and accessing appropriate healthcare support. Rare diseases such as CMT give rise to a number of medical and social issues. Due to a lack of awareness and funding, diagnostic delay occurs frequently and access to medical and social care is often unsatisfactory (Schieppati et al., 2008). The current study suggests that
awareness of, and support for CMT is generally limited, and this adds to the burden of CMT on HRQoL. Despite being generally inadequate, where awareness of CMT and healthcare support did exist it was highly valued by participants and perceived to positively influence HRQoL.

Lack of awareness and knowledge about CMT made disclosure to others challenging. Disclosure of CMT often occurred within the context of minimal or no understanding about CMT and this caused additional complications for support seeking. These challenges were compounded by the fact that, for many people, the condition is largely invisible. Thus, others are often not able to understand the challenges people with CMT face because they are not aware of the condition and they cannot observe the impact it has. Whilst some participants chose to make the invisible visible and found this beneficial, many participants did not want to draw attention to their condition. The unknown, invisible nature of CMT has been previously discussed within the qualitative literature (Lafarge et al., 2014; Arnold, McEntagart and Younger, 2005) but has been overlooked within quantitative studies. Disclosure of CMT is complex, in part due to the lack of awareness and the invisible nature of the condition. Previous research with other chronic conditions has indicated that rare and invisible conditions are associated with perceived isolation (Brennan and Creaven, 2016). Increased awareness of CMT may enable individuals living with the condition to receive...
a timely diagnosis and access specialist healthcare. It may also help people to feel better able to share their diagnosis with others.

Individually, and in combination, “maintaining independence: adaption, adjustment and support” and “awareness of CMT” had the potential to positively and negatively influence HRQoL. In order to overcome the every-day challenges and lack of control experienced, participants sought to maintain their independence via continual adjustments and adaption and by seeking support. Whilst this process had the potential to positively influence HRQoL, it sometimes adversely affected participants sense of identity and self-esteem and therefore detrimentally affected HRQoL. Maintaining independence was facilitated by improved awareness of CMT, and vice versa, however, in general, awareness of CMT was lacking. This lack of awareness exacerbated anxiety and threats to identity and self-esteem. It also prevented participants from seeking appropriate support and feeling comfortable disclosing their condition to others.

4.2.3 Positive influences. The final theme “resilience and positive growth” captured the way in which participants overcame the challenges they were faced with to re-define and enhance their perceived HRQoL. Previous research has largely overlooked positive psychological aspects of HRQoL, with a strong emphasis placed on negative psychological outcomes (e.g. depression and anxiety) and a deficit model of
functioning (e.g. see Johnson et al., 2014). Whilst a general link between resilience and quality of life is well-established (Alriksson-Schmidt, Wallandar and Biasini, 2007) and resilience has been identified as an important aspect of HRQoL in other health conditions (Nightingale et al., 2011), it has only been identified in one previous qualitative study in adults with CMT (Lafarge et al., 2014). Consequently, limited attention has been paid to this important construct thus far. The current study suggests resilience, the ability to bounce back from adversity (Smith et al., 2008), may be an important determinant of HRQoL in individuals with CMT. This may be due to the fact it helps people to maintain valued activities and roles in the face of every-day challenges.

Participants in the present study also illustrated many examples of positive growth such as finding benefits in their experience, making self-enhancing social comparisons, and expressing gratitude for the abilities and experiences they retained, and for their relationships with important others. Gratitude is a difficult concept to define but literally translates as ‘gratefulness’. Limited previous research has identified gratitude as an important emerging concept within HRQoL (Virani, 2017). Although not apparent in previous qualitative and quantitative CMT research, an experimental study of a brief gratitude intervention for adults with neuromuscular disorders reported significant improvements in subjective well-being (Emmons and McCullough, 2003). Thus, the current study extends previous knowledge and highlights the positive influence of gratitude on HRQoL in adults with CMT.
Despite evidence to suggest that benefit finding and quality of life are not related (Helgeson, Reynolds and Tomich, 2006), the current study suggests this may be an important psychological process in adults with CMT which positively influences HRQoL. Findings from the current study align with Cognitive Adaption Theory (Shelley, 1983) which suggests that cognitive adaption to threatening events involves three key elements: a search for meaning, an attempt to regain mastery, and restoring self-esteem through self-enhancing evaluations. Accordingly, participants in the present study identified meaning within their experiences and benefits they had gained from living with CMT. They also found new ways to gain a sense of mastery over valued activities and made self-enhancing social comparisons with others living with CMT and those with other health conditions. Given the importance of cognitive adaption for HRQoL (Livneh, 2001), these processes may be useful to further investigate in both research and practice.

“Resilience and positive growth” therefore positively influenced HRQoL. There were reciprocal relationships with “awareness of CMT” and “maintaining independence: adaption, adjustment and support”. For example, support for CMT, where it existed, helped to enhance positive growth and resilience. Additionally, positive growth and resilience motivated participants to raise awareness of CMT.
4.3 Theoretical frameworks

Broadly speaking, the themes identified within the preceding analysis align with Wilson and Cleary’s (1995) conceptual model of HRQoL. This identified the inter-connected concepts of biological factors; symptoms; functioning; health perceptions; emotional and psychological factors; characteristics of the environment; and overall quality of life. Accordingly, a range of physical symptoms and functional impairments were perceived to influence HRQoL, however these were experienced within a broad and individualised context of internal and external factors. Psychologically, a number of factors facilitated or impaired HRQoL. Anxiety, low sense of control, low coping self-efficacy and impaired self-concepts were key barriers, whereas finding positives and meaning, experiencing gratitude and positive growth were key facilitators. The environmental context in which CMT is experienced is extensive and complex. It includes, for example, the support participants received from friends and family, Government backed mobility assistance schemes, access to Healthcare services and physically and socially restrictive environments. Since many of these aspects are currently under threat (e.g. Mobility assistance schemes) or are insufficient (access to Healthcare service), the environmental context represents a key barrier to HRQoL. Despite much adversity, in accordance with the ‘Response-Shift’ model (Sprangers and Schwartz, 1999), participants’ frequently found new ways to achieve their goals and/or re-prioritised and re-conceptualised them. Thus, overall HRQoL was sometimes preserved, in spite of the substantial physical burden of CMT.
Across participants, the importance of maintaining independence and autonomy was evident, as was the importance of successfully engaging in valued activities and roles and maintaining connections with others. In addition to clear links with the Disability Centrality Model (Bishop, 2005) which highlights the salience of valued activities and roles, these findings allude to the importance of self-determination. According to self-determination theory (Deci & Ryan, 2000; Ryan & Deci, 2000), three core conditions are necessary for positive functioning: autonomy, competence and relatedness. Thus, in order to achieve a good HRQoL, these conditions must be met (Zubialde, Mold and Eubank, 2009). Self-determination theory may therefore offer a useful framework for understanding the psychological aspects of HRQoL in adults with CMT. Despite its clear applicability to the current findings, previous CMT studies have not made links to this theory. This is therefore an important new finding, which may have been facilitated by the novel photovoice method which provided participants with considerable autonomy over how they portrayed their experiences. Indeed previous photovoice investigations have frequently identified new insights, beyond those achieved via traditional qualitative methods (e.g. Williams, Sheffield and Knibb, 2016; Aubeeluck and Buchanan, 2006; López and Randall-David, 2005). Self-determination has previously been argued to be of particular relevance to individuals with chronic health conditions, with important implications for how HRQoL should be assessed (Zubialde, Mold and Eubank, 2009).
Following the initial analysis, an overall framework for understanding HRQoL in individuals with CMT was developed and is provided in Figure 2. This highlights interconnected physical, social and psychological components, in addition to condition management and independence. Internal (e.g. values/ preferences) and external (e.g. awareness of CMT, accessibility, funding for healthcare and disability support) factors influence these components and are therefore crucial in understanding HRQoL.
Social support and connection with others
Engagement in valued roles (e.g. work, parent etc.)

Physical
Symptoms (e.g. reduced sensation, muscle weakness, poor balance, structural changes to feet and hands, pain, fatigue etc.)
Functioning (e.g. limited mobility, altered gait, reduced strength & dexterity, falls, difficulty climbing stairs etc.)
Progression of CMT symptoms and impact on functioning

Internal factors: Individual values and preferences

Psychological
Autonomy
Resilience
Acceptance and positive growth
Identity
Self-esteem
Appearance concerns
Perceived control over condition
Future-orientated anxiety

CMT management
Access to & satisfaction with specialist healthcare and adaptations/orthotic devices to support condition management (e.g. podiatry/chiropody, physiotherapy, surgery etc.)
Self-management (e.g. adapted exercise, foot care)

Independence
Engagement in valued activities
Mobility/disability assistance

Figure 2: HRQoL Framework
4.4 Implications for HRQoL Assessment

Findings from the current study have a number of implications for the assessment of HRQoL in adults with CMT. Similar to the current study, previous qualitative research has identified that aspects of the environment, social relationships and psychological factors are often discussed by patients with neuromuscular disorders to a greater extent than the physical aspects of their condition (Dany et al., 2017). Despite this finding, there is often a disconnect in HRQoL assessment development which typically adopts a symptom-based approach. This adds to the general concern that measures of HRQoL are not patient-centred and therefore may not be valid or sensitive to change (Carr and Higginson, 2001).

Existing measures of HRQoL may not fully capture the complexity of this individualised, multi-faceted construct for adults with CMT. There is widespread agreement that generic scales such as the SF-36 (Ware and Sherbourne, 2012) are not adequate for people with CMT (McCorquodale, Pucillo and Johnson, 2016, Burns et al., 2012). Importantly, this measure does not include items related to perceptions about the future, self-perceptions, independence and relationships which have previously been argued to be important to individuals with neuromuscular conditions (Carr and Higginson, 2001), and have been identified in the current study. Following a call for more specific HRQoL measures (e.g. Burns et al., 2012), the ‘QoL-NMD’ scale was recently developed to measure HRQoL in individuals with slowly progressive neuromuscular conditions (Danny et al., 2015). Although CMT fits within this sub-type
of neuromuscular conditions, individuals with CMT do not appear to have been included as participants in the development of this scale. The scale consists of three domains: (1) impact of physical symptoms, (2) self-perception and (3) activities and social participation. Items were removed from the scale if a percentage of participants did not respond to the question, or indicated it was not important. Unfortunately, the current findings indicate a number of these removed items may be particularly relevant for individuals with CMT such as disability allowance, access to public transport, access to employment, contact with patient organisations and items relating to starting a family in the past or future. Thus, this scale may not be appropriate for individuals with CMT since potentially important aspects of HRQoL may be overlooked.

Of the limited number of patient-reported outcomes developed specifically for individuals with CMT, one is a measure of neuropathy (CMTNS) (Shy et al., 2005) and one is a measure of disability (mDSI) (Ramchandren et al., 2014), as defined according to the medical model of disability which emphasises impairment. Thus, symptoms and functioning have been prioritised in patient-reported outcomes specific to CMT.

Although one previous study has reported a quantitative survey of items pertaining to HRQoL in adults with CMT (Johnson et al., 2014), this has similarly adopted a symptoms-based approach. An extensive number of items were included in this survey, arguably beyond the standard concept of symptoms as a patient’s perception of abnormality in physical, psychological or cognitive states (Wilson and Cleary, 1995). Whilst this scale may therefore go some way towards assessing HRQoL, some important concepts identified in the current study have been omitted, such as sense of
identity, access to disability benefits and acceptance. Furthermore, the scale asks respondents to evaluate the impact of each symptom on their life but does not ask about relative importance.

The current study suggests that different aspects of HRQoL are differentially important to individuals. This is consistent with Wilson and Cleary’s (1995) conceptual model of HRQoL, which emphasises the role of patient preferences and values. Thus, any assessment of HRQoL for adults with CMT should ensure that perceived importance of items is considered, in addition to perceived impact. Relative importance of aspects of HRQoL may be influenced by an individual’s sociodemographic variables (e.g. marital status, socioeconomic status, geographical location, age, sex), individual and cultural belief systems and expectations and aspirations (Carr and Higginson, 2001). In order to appropriately capture importance, individuals should be asked to weight the relative importance of HRQoL items (Carr and Higginson, 2001). Individually generated measures may also be advantageous. For example, a similar approach to the Patient Generated Index (Ruta, Garratt and Russell, 1999) could be adopted. The Patient Generated Index asks individuals to identify areas of their life which are affected by their health condition, to rate the severity of impact of these areas and then to identify areas they would want to improve if they could, using a point-based system to indicate relative importance. The authors suggest the final score should identify HRQoL in areas which individuals would most value improvement (Ruta, Garratt and Russell, 1999).
The concept of ‘improvement’ is indicative of the deficit model approach to HRQoL which predominates existing measures. Assessment of HRQoL typically focuses on negative symptoms and impairments in physical, psychological and social functioning (e.g. see Guérin, 2012). An absence of impairment is not necessarily synonymous with the way in which adults with CMT conceptualise HRQoL, however. Instead, as previously discussed, positive HRQoL for individuals with CMT can be understood as the ability to accept, adapt to and grow from the broad impact of CMT, with appropriate support, in order to maintain individually valued activities, roles, and identities, alongside valued connections with others. Thus, HRQoL assessment should include aspects of positive functioning, rather than relying solely on assessment of deficits. This supports Wilson and Cleary’s (1995) suggestion that HRQoL assessment should include some measure of satisfaction. As an example, HRQoL assessment could usefully consider whether individuals with CMT are satisfied with their relationships and/or feel their relationships are flourishing, as opposed to whether they perceive them to be impaired. Additionally, whilst HRQoL measures frequently assess psychological dysfunction (e.g. anxiety, depression); the current study suggests that positive psychological functioning should also be assessed. Whilst this could include specific traits such as resilience and gratitude, it may also be useful to consider broad domains of positive functioning linked to self-determination theory, such as perceived autonomy, social relatedness and competence (Deci & Ryan, 2000; Ryan & Deci, 2000).
A final implication of the findings in relation to assessment concerns the dynamic nature of HRQoL. Day to day fluctuations in the physical manifestations of CMT, in addition to the changeable social and environmental context, mean that HRQoL is inherently variable and at any point in time, different facets may be more or less important to individuals. Thus, in addition to assessment of the relative importance of variables, multiple assessment points may be preferable to one off measurements of HRQoL. Moreover, as acceptance develops, individuals may come to re-conceptualise HRQoL. As identified in the ‘Response-Shift’ model (Sprangers and Schwartz, 1999), participants’ in the present study found new ways to achieve their goals and/or re-prioritised and re-conceptualised them. Thus, HRQoL assessment should include, or be accompanied by, measures of acceptance of CMT. Alongside the other recommendations put forward, this would enable researchers and clinicians to better capture whether change in HRQoL is due to change in CMT-related variables, change in the social and environmental context, or conversely, change in acceptance of this complex condition. This may be particularly important when HRQoL is assessed as an outcome of interventions.

4.5 Interventions to improve HRQoL

A number of recommendations can be suggested with respect to interventions and HRQoL in adults with CMT. Given the complexity of HRQoL and its diverse influences, a variety of ‘interventions’ have the potential to improve HRQoL. These may
be best categorised according to the Determinants of Health model (Dahlgreen and Whitehead, 1991), which illustrates the different layers of influence on health and health outcomes.

At the level of individual biological factors, future treatments targeting gene expression may have the potential to dramatically improve HRQoL. Duplication of the \textit{PMP22} gene has been identified as the genetic cause of CMT Type 1A. Very recent trials have indicated that antisense oligonucleotides (ASOs)\textsuperscript{5} can stop the overproduction of \textit{PMP22} mRNA in rodent models. Furthermore, treatment with ASOs reversed the severity of pre-existing neuropathy and increased myelination in demyelinated neurons (Zhao \textit{et al.}, 2017). This could potentially have very significant benefits for those with CMT Type 1A, including a reduction in anxiety about progression and the risk of inheritance identified in the current study. Research is clearly still at a very early stage and caution must be exercised since promising potential treatments have not historically generalised from rodent models to human populations (e.g. trials with ascorbic acid) (Pareyson \textit{et al.}, 2011). Moreover, although CMT Type 1A is the most common form of CMT, it is not the only type and, as the current study indicates, HRQoL is impaired across the different forms of CMT. Furthermore, various factors influence perceived HRQoL, beyond the physical

\textsuperscript{5} Antisense Oligonucleotides (ASOs) = synthetic single-stranded nucleic acids which that bind to targeted mRNA to break it down.
manifestations of CMT. Thus, it is important that additional interventions are also implemented.

At the individual lifestyle level, adapted exercises may positively influence HRQoL, by maintaining functioning, slowing progression and/or improving a sense of control and self-efficacy. Indeed, many participants in the current study discussed the positive impact that exercise had on their HRQoL. Evidence from the current study and previous research indicates that exercise can improve perceived functioning and self-efficacy (McAuley and Blissmer, 2000). Additionally, and perhaps concurrently, psychological interventions designed to help individuals accept or change their relationship with anxious thoughts and/or build resilience and self-determination could be beneficial. These include psychological therapies such as Mindfulness-Based Cognitive Therapy (MBCT), and Acceptance and Commitment Therapy (ACT), in addition to positive psychology interventions such as Gratitude Journals. Whilst some therapeutic interventions will be reliant on appropriate healthcare access, some psychological interventions can be accessed individually. For example, Gratitude Journals do not require healthcare assistance and show promising benefits for individuals with CMT and other neuromuscular disorders (Emmons and McCullough, 2003).

At the level of social and community networks, peer-support interventions and access to CMT support organisations may also improve HRQoL. These could potentially be delivered online due to the relatively low numbers of individuals living with CMT,
and the physical burdens of the condition. Previous research has indicated online support interventions significantly increase social networking and are valued by young people with disabilities (Raghavendra et al., 2013). Although concerns exist about accessibility of digital interventions for older populations, the number of older adults accessing the internet is steadily increasing (Green and Rossall, 2013). Furthermore, a number of older adults successfully took part in this digital photovoice study. Therefore, digital information and social support may be an important intervention to improve HRQoL in this group.

It is important to recognise that intervention opportunities exist beyond the social and community level which directly and indirectly influence HRQoL. Crucially, a number of intervention opportunities occur at the living and working conditions level. Increased awareness and healthcare support for CMT is required to facilitate specialist physiotherapy, occupational therapy, orthotics, surgical intervention, psychological therapy and genetic counselling, as needed. Ideally, healthcare should be provided in multi-disciplinary teams. Importantly, healthcare professionals must acknowledge that interventions may not automatically improve HRQoL for every individual. For example, orthotics may improve walking ability but may cause pain, restrict footwear and clothing, and detrimentally affect women’s femininity. Thus, an individualised, patient-centred approach to the delivery of healthcare is vital. This recommendation is supported by a recently published systematic review which concluded that patient preferences must be incorporated into clinical decision making to improve outcomes for individuals with neuromuscular disease (Landfeldt et al., 2017). In addition to
healthcare access and delivery, practical support to help adults with CMT remain in full-time or part-time employment may also benefit HRQoL. Where remaining in employment is no longer possible, supportive counselling should be offered to help individuals resolve threats to their sense of identity and self-esteem. As with other forms of psychotherapy, this should be underpinned by the central components of self-determination theory and support the individual’s basic psychological needs for autonomy, competence and relatedness (Ryan and Deci, 2008).

Finally, vital opportunities for intervention exist at the level of general socio-economic, cultural and environmental conditions. Again, these have the potential to both directly influence HRQoL, and to influence access to interventions at the lower levels. Government funding to support research and to make improvements in disabled access, disability benefits and access to long-term (as opposed to time-limited) healthcare is crucial. Moreover, National Institute for Health and Clinical Excellence [NICE] guidelines are urgently required to improve diagnosis, assessment and management of CMT. Such guidance has the potential to standardise best practice across the UK and, consequently, to substantially improve HRQoL.

4.6 Strengths and limitations of the current study

The photovoice method employed within the current study provided a unique, in-depth insight into the every-day nuances of health-related quality of life, as it is
experienced by adults living with CMT. In combination, the illustrations and textual reflections provided a window into participants’ experiences and the meanings they attached to HRQoL. Arguably, the richness of the data within the present study may not have been gathered using more traditional qualitative methods such as interviews. Importantly, the incorporation of participant-generated photographs and written narratives enabled participants to express themselves using multiple modalities, thus reflecting the multi-modal nature of experience (Reavey and Prosser, 2012). Furthermore, the two-week time period of participation enabled participants to illustrate and describe the dynamics of HRQoL over time. It is reasonable to assume that one-off interviews may be influenced by the way a participant is feeling at that particular point in time. As such, capturing experience over time may be preferable in order to identify key themes which usefully characterise complex, multi-faceted concepts such as HRQoL.

The minimal direction imposed by the researcher allowed for participants to determine what they considered to be important aspects of HRQoL and to communicate this in a way which was meaningful for them. In accordance with Williams, Sheffield and Knibb (2016), no definition of HRQoL/QoL was provided to participants. This is because one of the aims of the study was to understand how HRQoL was perceived by individuals with CMT. Providing a standard definition may have restricted responses to those which conformed to a pre-imposed
conceptualisation of HRQoL. By extension, this may have led to potentially important insights being overlooked (Williams, Sheffield and Knibb, 2016). As a consequence of the minimal direction provided by the researcher, participants varied in the number of journal entries they posted and the approach taken to individual posts. Some participants completed the study as a daily diary, with photographs and reflections taken in response to experiences which occurred on a given day. Others identified a key theme for each entry and submitted photographs and reflections to illustrate these themes. Whichever approach participants took, similar issues and experiences emerged from the journal entries. This was profoundly illustrated during one critical incident where two participants discussed the importance and value attached to possession of a disabled badge in separate journal entries posted on the same day. One participant was following a daily diary approach and the other structured the journal according to key themes (see Appendix C II for reflective diary entry). The flexibility afforded to participants can therefore be viewed as a key strength of the study since participants were actively involved in choosing how they engaged with the method and the content they would share with the researcher. Conversely, it could be contended that the minimal direction is also a limitation of the study. Providing maximal control to participants means that there was divergence in the frequency and number of journal entries, as well as the included content. Moreover, a number of participants included some photographs which could not be meaningfully and/or ethically included in this thesis e.g. photographs of identifiable children/ adults and photographs which did not appear to have been participant generated (including a
photograph of a bus and a photograph of a swimming pool). Whilst providing stricter
guidance may have increased comparability between participants, divergence within
the current study did not prevent the identification of key themes across participants’
visual and textual narratives.

It is conceivable that the process of taking photographs and explaining their
meaning encouraged participants to reflect on their experiences in depth, and assisted
in developing meanings. This concept of the method aiding reflection was highlighted
during one participant’s first journal entry (see Appendix C II for example reflective
diary entry). A brief initial textual description was subsequently added to with
poignant detailed reflection about what the image represented. In addition to
highlighting the value of accompanying textual narratives for interpreting visual
images, this critical incident highlighted the reflective process of the method itself. It is
reasonable to conclude that the photovoice method employed in the current study
may have helped participants to make sense of their HRQoL and the factors which
influence it.

The novel online photovoice method gave rise to a number of strengths and
limitations. Key strengths included accessibility, control and the ability to document
and reflect upon experience in real-time. Despite the researcher’s initial concerns that
the method may lead to a biased sample of younger adults, these concerns were
unfounded. Indeed two participants were in their 70’s at the time of participation. For individuals with limited mobility, it is likely that the option to send photographs to the researcher via a mobile phone or home computer meant that the study was less burdensome than traditional methods. Furthermore, participants were not required to travel to meet the researcher and discuss their photographs. Digital photographs enabled participants to select photos to upload and removed the risk that some photos would not develop, a limitation reported in previous studies (Williams, Sheffield and Knibb, 2016). Participants also had the option to add to or amend their posts if they wished to during the study period which further enhanced autonomy and control. Additionally, the remoteness of the researcher may have helped some participants to photograph and discuss experiences freely that may have been difficult to discuss within a face to face context. In addition to enhanced control over the material presented, participants were able to upload photographs and textual descriptions as experiences occurred. It is likely this ability circumvented issues reported in other photovoice studies where participants were unable to recall meaning behind some of the photos they had taken (Thompson et al., 2008).

A number of limitations with the digital photovoice method must also be acknowledged. Unlike face to face interview methods, the researcher was not able to clarify meanings within the textual descriptions of photographs. As such, areas of importance could have been overlooked or, conversely, heightened. This limitation is not unique to a digital approach but rather is specific to journal/diary methods. Unlike
some previous photovoice studies (e.g. Lockett, Willis and Edwards, 2005), participants were not asked to select their most meaningful photographs. Instead, all photographs and textual descriptions were assumed to be meaningful. This again raises the possibility that importance could have been over-stated in some areas. This is considered unlikely, however, since themes were developed across participants and across visual and verbal data.

An important consideration when interpreting the findings from the present study concerns the participant sample and sampling strategy. Although participants did not need to be members of CMT-UK to see the study advert, non-members did need to follow the Facebook page. Therefore participants who took part in this study may have been particularly well informed about their condition and likely to find support helpful. Previous research in this field has often recruited participants from clinic registers (e.g. Johnson et al., 2013) however, patient support organisations have also been accessed (e.g. Lafarge et al. 2014; Alberts, 2009; Arnold, McEntagart and Younger, 2005) and may be advantageous due to the high number of individuals with CMT who are not in receipt of specialist healthcare. Multi-site recruitment (e.g. clinic registers and patient support organisations) may be beneficial in future research if generalization is sought.
Although there was considerable heterogeneity in CMT type and self-reported functioning, the mean age of participants was relatively high. Furthermore, the majority of participants who took part in the study were female. This may mean that the perspectives of younger adults and males were less well represented. Indeed, despite a clearly identifiable impact of CMT on femininity, the impact on masculinity was less evident. Given that some previous quantitative research has identified greater impairments in HRQoL in women and in older adults (Johnson et al., 2014; Redmond, Burns and Ouvrier, 2008; Vinci et al., 2005), it could be that participants in this study were particularly likely to experience adverse HRQoL. Whilst not all studies support these differences (Taniguchi et al., 2013; Redmond, Burns and Ouvrier, 2008; Padua et al., 2006), and the current study did not seek to identify generalisable findings, future research may benefit from utilising theoretical and purposive sampling techniques to ensure adequate representativeness across key demographic and disease indicators.

A further limitation to the current study relates to attrition. Five participants consented to take part but never accessed their online journal and/or never submitted a journal entry. Thus, although a diverse age demographic was obtained, it is possible that only individuals who were particularly comfortable with digital technology participated. Previous photovoice studies have frequently reported high levels of attrition and have recommended digital methods to overcome this issue (e.g. Williams, Sheffield and Knibb, 2016). The current study suggests that attrition is also an issue for
digital methods and typically occurs between consent and participation. Although life events were reasons for non-participation in some cases, most participants who did not start the study did not provide a reason. It may be that greater participant-researcher interaction at the start of the study could help to reduce attrition. Thus, future studies may benefit from including a telephone or face to face discussion at the start of the study for all participants. This could potentially include some form of photovoice training. There is some debate in the literature as to whether photovoice training could potentially constrain and alter the way participants choose to represent their experiences (Topcu, 2015). Despite this concern, training is common in the majority photovoice studies and has been found to lead to increased engagement (Catalani & Minkler, 2010). Hence, future research using a digital photovoice method could look at ways to incorporate participant training in photovoice. It is possible that some participants may have experienced technical difficulties which led them to cease participation. A small number of participants who completed the study reported experiencing a technical difficulty at some point during the study’s duration. It is unlikely that this was a major cause of attrition, however, since most participants who dropped out never logged on to the journal area. Furthermore, comprehensive written and visual guidance was provided to all participants about how to access and use the journal. An alternative explanation is that the study was considered overly burdensome and time consuming. Future studies may benefit from a shorter time period of participation (e.g. one week) to reduce participant burden, whilst still providing the opportunity to capture experience over time.
A final strength and limitation concerns my own lived experience of CMT. Having shared experience can be viewed as a strength since it enables valuable insight and knowledge to underpin the research (Berger, 2013). This insight and knowledge was enhanced by working with a patient partner throughout the study duration. Thus, the study was conducted from an informed perspective. Whilst such insight can be a key strength, it may also represent a potential limitation. It is important to acknowledge that researchers are active within the analytic process (Braun, Clarke and Terry, 2015). Thus, although I had minimal influence on what participants chose to photograph and write about, the final analysis is inevitably influenced by my own experiences of CMT which will have shaped how I interpreted and engaged with the data. As can be seen from my initial reflection (Appendix C I), my own experiences and beliefs about HRQoL generally map to those of the participants of this study and the final themes. It is important to note that some degree of influence on the analysis is true of all qualitative researchers, irrespective of shared experience. Furthermore, I followed available guidance for reducing the risks associated with shared experience (Berger, 2013).
4.7 Directions for future research

In addition to the suggestions made above to address the limitations to the present study, a number of important directions for future research can be suggested. Firstly, given its successful implementation in the current study, the digital photovoice method employed within the current study could be usefully applied to a range of health conditions associated with impaired mobility. Within the field of CMT research, future studies could use the digital photovoice method to examine HRQoL in adolescents and young adults with CMT, who may understand this dynamic concept in a different way to participants in the present study. Additionally, further qualitative research could explore the concepts of masculinity and femininity in more depth within CMT populations.

 Crucially, future qualitative and quantitative research should further investigate the new and overlooked aspects of HRQoL identified in this study, such as access to healthcare, physical activity, identity, stress, resilience and self-determination. In addition to cross-sectional and longitudinal studies designed to assess the predictive value of these factors, controlled trials of interventions aimed at improving HRQoL are urgently required. Interventions should be developed in consultation with individuals with CMT and focus on addressing identified determinants of HRQoL.
In order for the benefits of interventions to be validly assessed, it is important that research proceeds to the development and psychometric testing of a CMT-specific measure of HRQoL, rather than continuing to use inappropriate general measures. This should not be limited to physical symptoms and functioning, but instead should incorporate broader aspects of HRQoL and individual values. Moreover, it should include positive aspects of HRQoL which are frequently overlooked in existing scales. As HRQoL is inherently complex, however, mixed-method evaluations may always be preferable. Development of a CMT-specific HRQoL measure should not, therefore, displace qualitative research, since capturing the dynamics of HRQoL may always be difficult to attain with quantitative measures.

4.8 Conclusion

To conclude, this novel digital photovoice investigation adds to the limited body of literature related to HRQoL in adults with CMT. Participants in the present study illustrated and discussed aspects of HRQoL which are personally meaningful and important to them. HRQoL is dynamic and individual but may be tentatively conceptualised as the ability to accept and adapt to the physical aspects of CMT, with support, in order to maintain independence and engage in valued roles and activities, to sustain relationships with important others and to attain positive growth. Individuals with CMT experience daily challenges and barriers to attaining HRQoL, across the broad determinants of health. Thus, whilst the physical manifestations of CMT are
numerous and inherently aversive, psychological, social, environmental and socio-political factors all play an important, nuanced role in HRQoL. Increased awareness of CMT, the provision of specialist healthcare support backed by NICE guidance, and access to mobility assistance schemes are vital to reduce the detrimental impact of this complex condition on HRQoL. Furthermore, interventions aimed at improving HRQoL must be urgently developed and evaluated. To improve the rigour and validity of evaluative research, and to improve outcome monitoring in routine clinical practice, it is imperative that research proceeds in the development and testing of a CMT-specific measure of HRQoL. Developments in research and practice should continue in partnership with individuals living with CMT, who are invariably best placed to advise on the broad impact of this condition. Crucially, to positively influence HRQoL, research, policy and practice must seek to enhance the basic psychological needs of adults with CMT, and recognise the importance of individual values.
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Appendix A - Systematic Review

The following systematic review was submitted for USPJKH-30-M. It has since been published in an open-access journal:


Do Psychosocial Interventions Improve Quality of Life and Wellbeing in Adults with Neuromuscular Disorders? A Systematic Review and Narrative Synthesis.

**Background**: The objective of this paper was to systematically review and critically appraise all known empirical studies of psychosocial interventions designed to improve quality of life and well-being in adults with neuromuscular disorders. **Method**: A systematic review of the published and unpublished literature was conducted. Studies meeting inclusion criteria were appraised using a validated quality assessment tool and results presented in a narrative synthesis. **Findings**: Out of 3,136 studies identified, ten studies met criteria for inclusion within the review. Included studies comprised a range of interventions including: cognitive behavioural therapy, dignity therapy, hypnosis,
expressive writing, gratitude lists, group psychoeducation and psychologically informed rehabilitation. Across varied interventions and neuromuscular disorders, seven studies reported a short-term beneficial effect of intervention on quality of life and well-being. Whilst such findings are encouraging, widespread issues with the methodological quality of these studies significantly compromised the results. **Discussion:** There is no strong evidence that psychosocial interventions improve quality of life and well-being in adults with neuromuscular disorders, due to a paucity of high quality research in this field. Multi-site, randomised controlled trials with active controls, standardised outcome measurement and longer-term follow-ups are urgently required.

Neuromuscular disorders are a heterogeneous group of acquired and inherited conditions affecting the neuromuscular system (Sandoval, 2006). There are four main categories of neuromuscular disorders, namely motor neuron disorders (e.g. Amyotrophic Lateral Sclerosis [ALS]), nerve junction disorders (e.g. Myasthenia Gravis), muscle disorders (e.g. Duchenne Muscular Dystrophy) and peripheral nerve disorders (e.g. Charcot-Marie-Tooth disease) (Hilton-Jones, Freebody and Stein, 2011). It is estimated that 70,000 people live with some form of neuromuscular condition in the UK (Muscular Dystrophy Campaign, 2010). Of this figure, approximately 22,750 are estimated to be adults living with a severe and chronic neuromuscular condition (Hill and Phillips, 2006). Concerns have been repeatedly raised about the lack of appropriate service provisions to address the complex needs of adults with
neuromuscular conditions (All Party Parliamentary Group for Muscular Dystrophy [APPGMD] 2009, 2015; Hill and Phillips, 2006). Although varied in clinical presentations, all neuromuscular conditions cause degeneration of the nerves or muscles controlling sensation and/or movement and most are progressive and incurable (Schepelmann et al., 2010). The physical impact on the individual is often substantial and includes muscle weakness, fatigue, pain, restricted activities of daily living and numerous secondary health complications, leading to a significantly reduced life-span in some conditions (Van der Beek, 2013; Pagnini, 2012; Sadoval, 2006). As a consequence, neuromuscular disorders are associated with many adverse psychosocial outcomes, such as reduced quality of life and well-being (Vinci et al., 2005; Piccininni, Falsini and Pizzi, 2004).

The World Health Organisation (WHO) defines quality of life as the way in which an individual perceives their individual position in life with respect to their goals and standards and suggests this broad construct can be affected by the individual’s physical health, psychological health, independence, and relationships with others and the environment (WHOQOL Group, 1995). Health-related quality of life (HRQoL) is one aspect of this multi-faceted concept and refers to the physical, social and psychological domains of health, influenced by an individual’s beliefs, expectations and experiences (Testa and Simonson, 1996). Despite clinical variation between slowly progressive (e.g. Facioscapulohumeral Muscular Dystrophy) and rapidly progressive (e.g. Amyotrophic Lateral Sclerosis) neuromuscular conditions, perceptions of self-reported HRQoL have
been found to be comparably low in individuals with both categories of disorders (Özer et al., 2010).

Recently, researchers have noted that quality of life assessments typically focus on *deficits* in functioning and have argued that an absence of deficits is not synonymous with positive functioning. Consequently, it has been suggested that well-being, an *asset* approach to functioning assessing positive psychological variables, should also be considered as an important outcome in chronic illness (Upton and Upton, 2015; Guérin, 2012). Similarities in definitions of well-being and quality of life have led to confusion in the literature, with both terms often used interchangeably (Camfield and Skevington, 2008). Arguably a difficult construct to define, one key conceptualisation of well-being, subjective well-being, is associated with affect and life satisfaction. It has been described as ‘an umbrella term for different valuations that people make regarding their lives, the events happening to them, their bodies and minds, and the circumstances in which they live’ (Diener, 2006: 400). In contrast, psychological well-being encompasses concepts such as personal growth, mastery, resilience and acceptance (Burns and Ma, 2015). The importance of well-being in neuromuscular disorders is well-established. Indeed, research has found adults with amyotrophic lateral sclerosis and high well-being have a risk of mortality seven times lower than those experiencing distress, irrespective of disease severity and length of illness (McDonald et al., 1994).
Given the importance of both quality of life and wellbeing in the experience of
euromuscular disease, it is essential that effective interventions are identified.
Indeed, both patients with neuromuscular disorders and researchers have argued this
should be a research priority (Graham et al., 2015; Nierse et al., 2013). Understanding
factors which affect quality of life and well-being is important for the design and
delivery of effective interventions. Critically, research has consistently found that
psychosocial factors are better predictors of quality of life than physical impairment in
adults with neuromuscular disorders (Graham et al., 2014; Roach et al., 2009; Simmons
et al., 2000). Such findings indicate that interventions targeting psychosocial factors
could therefore improve quality of life and well-being in this group.

Psychosocial interventions are interventions which target psychological and/or
social factors, as opposed to biological factors. These include, for example, all
psychological therapies (e.g. cognitive behavioural therapy), psychoeducation and peer
support. Such interventions can be delivered in individual and group formats (Ruddy
and House, 2005). Systematic reviews and meta-analyses indicate these interventions
can improve quality of life and well-being in adults with long-term conditions such as
cancer and diabetes (Osborn, Demoncada and Feurerstein, 2006; Rhese and Pukrop,

To date, no systematic review has considered the effectiveness of psychosocial
interventions for improving quality of life and well-being across neuromuscular
disorders. This is problematic as the effectiveness of these interventions and
methodological quality of the research undertaken to assess effectiveness is largely unknown. Given that National Institute for Health and Care Excellence (NICE) Guidelines are currently being developed for uncommon neuromuscular conditions (APPGMD, 2015), it is timely that the evidence for psychosocial interventions in neuromuscular disorders is systematically reviewed to inform future recommendations. Thus far, one published systematic review has assessed psychotherapy and pharmacotherapy interventions for improving well-being and reducing distress in ALS. Based on the results of four included studies, the authors concluded that there was insufficient evidence to make recommendations for psychotherapy (Gould et al., 2015). It should be noted, however, that the focus of this review on one aspect of psychosocial interventions (psychotherapy) and a single neuromuscular condition limits the wider applicability of these findings. The importance of reviewing evidence across neuromuscular disorders for non-pharmacological interventions has previously been highlighted but, so far, has been limited to exercise and physical therapy interventions (e.g. Cup et al., 2007). A systematic review on the effectiveness of psychosocial interventions for improving quality of life and well-being in adults with neuromuscular conditions is now required.

Hence, this review sought to address gaps in the evidence and investigate the effectiveness of psychosocial interventions across neuromuscular disorders, with a view to making recommendations about effective interventions and developments for future research. There were two main aims: (1) To systematically review the literature on the impact of psychosocial interventions on quality of life and wellbeing in adults
with neuromuscular disorders; (2) To critically appraise the methodological quality of papers included in the review.

Method

Search Strategy

A systematic search of published and unpublished literature was conducted between June and October 2015. This included the following online databases: PsycInfo (1909-present), PsycArticles (1909–present), Medline (1964–present), CINAHL (1984-present), AMED (1988-present), Web of Science (1900 - present), and the Cochrane Register of Controlled Clinical Trials (CENTRAL). Databases were searched on 9th June 2015 using a Boolean search devised in PsycInfo and adapted to other databases (see Appendix A for full search strategies). Due to the small number of studies identified in initial scoping searches, a broad set of free-text search terms were devised around the key concepts of neuromuscular disorders, psychosocial interventions and quality of life/well-being. Search terms were reviewed and approved by a University Librarian and included:

1. ("neuromuscular disorder*" OR "neuromuscular*" OR "muscular dystrophy" OR "myasthenia Gravis" OR "charcot-marie-tooth" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome")
AND

2. (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR counseling OR "psychological therap*" OR psychotherapy OR psychodynamic OR "cognitive behavio#r*" OR cognitive OR behavio#r* OR CBT OR "self manag*" OR "self care" OR mindfulness OR "acceptance and commitment therapy" OR tele*)

AND

3. ("quality of life" OR "well being" OR "well-being" OR "wellbeing" OR "life satisfaction" OR "mental health" OR "psychological stress" OR anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "psychological adjustment")

A similar search strategy was employed to search COS Conference Papers Index and Open Grey. Hand searching of the Cochrane Neuromuscular Disease Group (all publications) was also conducted. To broaden the search, identified experts in the field and neuromuscular disease charities were also contacted. Citation tracking (using Google Scholar) and hand searching of reference lists were carried out for all included studies.

**Inclusion/exclusion criteria.** To be included in the current review, identified studies had to meet the following criteria:
1. Study design either randomised controlled trial (RCT), controlled trial or cohort study.

2. Participants all adults (18+) with a diagnosis of a neuromuscular disorder (or 75% with NMD diagnosis if sample includes mixed conditions). In accordance with previous reviews, patients with neuromuscular symptoms not attributed to a specific neuromuscular disorder and patients with diabetic neuropathy, entrapment neuropathy and radiculopathies were not included (Cup et al. 2007).

3. Participants received a psychosocial intervention (e.g. psychotherapy, psychoeducation etc.). In accordance with Ruddy and House (2005), multi-faceted interventions including a biological component but with a clear psychosocial emphasis were included.

4. Quality of life and/or well-being assessed as quantitative outcome measure.

5. Publication in English language.

Limits to date of publication were not applied since the aim of this paper was to systematically review all known empirical studies of psychosocial interventions designed to improve quality of life and well-being in this patient group.

**Study Selection**

To determine eligibility of identified studies, primary screening of titles and abstracts was conducted independently by two of the authors (EW & KM). Articles assessed as meeting the inclusion criteria were subsequently obtained in full-text and
subject to independent secondary screening by both authors. To facilitate secondary screening, a data extraction form was piloted with a subset of identified studies and developed for use across all studies (see Appendix B). In accordance with current guidelines, inclusion of studies was determined by agreement between both reviewers (Centre for Reviews and Dissemination 2009).

**Assessment of Quality**

The Effective Public Health Practice Project (EPHPP) Quality Assessment Tool for Quantitative Studies (Thomas *et al*., 2004) was utilised to assess methodological quality of all included studies. The EPHPP was deemed more appropriate for the current review than the Cochrane Risk of Bias Tool (Higgins and Altman, 2008) because the EPHPP was developed for use with all quantitative designs (not just RCTs) and has been found to have greater inter-rater agreement (Armijo-Olivo *et al*., 2012). The EPHPP tool allows reviewers to derive a global rating of quality from the following components: selection bias, study design, confounders, blinding and withdrawals and drop-outs. Quality of included studies was independently assessed by two of the authors (EW & KM), with disagreement resolved via discussion.

**Study Synthesis**

Due to study heterogeneity, a narrative synthesis was conducted. A narrative synthesis should involve a preliminary synthesis of the data, an exploration of

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relationships, assessment of quality of the evidence and the development of a theoretical model about the way in which an intervention works. Accordingly, quality appraisal, tabulation of the data, grouping by intervention type and study quality and analysis of moderator variables were used as tools to facilitate the current narrative synthesis (Popay et al., 2006). Although some studies included additional measured outcomes beyond quality of life and/or well-being, and a small number also included caregiver participants, this review focused specifically on outcomes pertaining to quality of life and/or well-being in adult patients with neuromuscular disorders.

Results

Included Studies

After removal of duplicates, a total of 3,136 studies were extracted from online databases and other sources. Following primary screening, 3,117 studies were excluded as ineligible, with common reasons for exclusion comprising: not an intervention study, not an appropriate sample (condition or age), not a relevant outcome and not an empirical study. Thus, nineteen articles were retrieved as full-text for secondary screening, of which ten articles were included within the review (see Appendix C for study selection flow diagram and reasons for exclusion of full-text articles). The high proportion of irrelevant articles retrieved from the full search indicates the search strategy had low specificity. However, there is always a trade-off between sensitivity and specificity, with the potential for missing important studies when specificity is high (Harden et al., 1999). Moreover, the results are in keeping with
a previous similar review in which a small number of studies were included from a broad search (Gould et al., 2015).

**Study Characteristics**

The vast majority of included studies (8 of the 10) were published in the last four years (see Table 1 for study characteristics). Of the included studies, three were randomised controlled trials (RCT) (Koopman et al., 2015; Van Groentijn et al., 2015; Averill et al., 2013), four were cohort analytic trials (CAT) (Kleinbub et al., 2015; Martínez et al., 2014; Ahlstöm et al., 2006; Emmons et al., 2003) and three were cohort (pre-post) designs (Aoun et al., 2015; Palmieri et al.; 2012; Boosman et al., 2011). Most studies were small, with the total number of participants ranging from eight – 80 ($M = 39.1$). Participant demographics were varied. Samples consisted of between 32% and 74% males. Half of the interventions were for participants with ALS (Aoun et al., 2015; Kleinbulb et al., 2015; Van Groentijn et al., 2015; Averill et al., 2013; Palmieri et al., 2012), the remainder were for participants with post-polio syndrome (Koopman et al., 2015), muscular dystrophies (Ahlstöm et al., 2006) and mixed neuromuscular conditions (Martínez et al., 2014; Boosman et al., 2011; Emmons et al., 2003). Interventions were varied and included cognitive behavioural therapy (Koopman et al., 2015; Van Groentijn et al., 2015; Martínez et al., 2014), hypnosis (Kleinbulb et al., 2015; Palmieri et al., 2012), dignity therapy (Aoun et al., 2015), expressive writing (Averill et al., 2013), gratitude lists (Emmons et al., 2003), comprehensive rehabilitation
(Ahlstöm et al., 2006) and a psychoeducational fatigue management group (Boosman et al., 2011). The length of interventions ranged from one hour to 10 days, delivered over periods of between one week and 18 months. Six of the interventions were home-based (Aoun et al., 2015; Kleinbulb et al., 2015; Martínez et al., 2014; Averill et al., 2013; Palmieri et al., 2012; Emmons et al., 2003) and four were delivered in clinic settings (Koopman et al., 2015; Van Groentijn et al., 2015; Boosman et al., 2011; Ahlstöm et al., 2006). The majority of interventions were face to face (Aoun et al., 2015; Kleinbulb et al., 2015; Koopman et al., 2015; Van Groentijn et al., 2015; Martínez et al., 2014; Palmieri et al., 2012; Boosman et al., 2011; Ahlstöm et al., 2006), with two delivered remotely via written instructions (Averill et al., 2013; Emmons et al., 2003), and one delivered online (Martínez et al., 2014). Outcomes pertaining to quality of life and well-being were measured in different ways across studies. Only four studies included a six month follow-up to investigate longer term effects of interventions (Kleinbulb et al., 2015; Koopman et al., 2015; Van Groentijn et al., 2015; Averill et al., 2013).
Table 1: *Intervention details and summary outcomes for included studies*

<table>
<thead>
<tr>
<th>Study</th>
<th>Study design</th>
<th>NMD((s))</th>
<th>No. of NMD p’s</th>
<th>Psychosocial Intervention</th>
<th>Content, context &amp; delivery</th>
<th>Duration</th>
<th>Control</th>
<th>QOL/well-being outcome: Measure(s)</th>
<th>Main findings for QOL and/or well-being</th>
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</thead>
<tbody>
<tr>
<td>Averill et al. 2013</td>
<td>RCT</td>
<td>ALS</td>
<td>Int: ((n = 24)) Con: ((n = 24))</td>
<td>Expressive disclosure</td>
<td>P’s asked to write or talk about the deepest feelings about their condition at home.</td>
<td>20 mins writing/talking for 3 days over 1 week</td>
<td>No intervention</td>
<td>PWB: Composite score derived from measure of QOL, affect &amp; depression</td>
<td>Significant increase in PWB in intervention group from baseline to 3 months but not maintained at 6 months. Significant decrease in PWB in control group from baseline to 3 months, not at 6 months.</td>
</tr>
<tr>
<td>Koopman et al. 2015</td>
<td>RCT</td>
<td>PPS (+ severe fatigue)</td>
<td>Int: ((n = 23)) Con: ((n = 22))</td>
<td>CBT</td>
<td>Face to face CBT tailored to fatigue in NMD delivered by CBT therapists in outpatient clinics using standard modules. Individual or with partner.</td>
<td>0 – 12 ((Mdn = 7)) 1 x hour sessions delivered over 16 weeks</td>
<td>Usual care</td>
<td>HRQoL: SF-36</td>
<td>No significant difference in HRQoL at baseline, end of intervention, 3 &amp; 6 month follow-up compared to control.</td>
</tr>
<tr>
<td>Author</td>
<td>Year</td>
<td>Design</td>
<td>Conditions</td>
<td>Intervention</td>
<td>Comparator</td>
<td>Usual care</td>
<td>Mental QOL</td>
<td>Notes</td>
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<tr>
<td>Van Groene stijn et al.</td>
<td>2015</td>
<td>RCT</td>
<td>ALS</td>
<td>Face to face CBT tailored to adjustment &amp; coping delivered by trained psychologists in outpatient clinics using standard modules. Individual or with partner.</td>
<td>Int: (n = 10)</td>
<td>5 - 10 1 x hour sessions delivered over 16 weeks</td>
<td>SF-36-MCS &amp; ALSAQ-40-EF (subscales)</td>
<td>QOL measured by ALSAQ-40-EF deteriorated less in the CBT group than control from baseline to 6 months. No significant change in SF-36-MCS.</td>
<td></td>
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<tr>
<td>Ahlstrom et al.</td>
<td>2006</td>
<td>CAT</td>
<td>MD: (MTD, FSHD, BD, LGD, ED, HDM, PD)</td>
<td>Individual and group sessions for patients and next of kin tailored to biopsychosocial needs. Delivered by counsellors, Occupational Therapists, Physiotherapists, neurologist &amp; nurse in hospital.</td>
<td>Int: (n = 37)</td>
<td>4 sessions delivered in 10 days over 18 month period</td>
<td>SIP (HRQoL), PWBQ (PWB) &amp; HADS (Mood)</td>
<td>No significant change in HRQoL, PWB or mood from pre to immediately post intervention. Control group deteriorated significantly on physical index of HRQoL.</td>
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<tr>
<td>Emmons et al.</td>
<td>2003</td>
<td>CAT</td>
<td>Mixed: PPS, CMT, FSHD</td>
<td>P’s wrote daily gratitude lists at home of up to 5 things grateful/thankful for &amp; Daily writing for 21 days</td>
<td>Int: (n = 33)</td>
<td>No intervention</td>
<td>SWB: Author developed scale. Observer reported well-being: SWLS &amp; author developed affect scale</td>
<td>Significantly higher SWB in intervention group than control across 21 day intervention period. Observer ratings significantly higher for</td>
<td></td>
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<tr>
<td>Study</td>
<td>Intervention Type</td>
<td>Conditions</td>
<td>Sample Size</td>
<td>Details</td>
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</tr>
<tr>
<td>Kleinbuet al.</td>
<td>Hypnosis-based</td>
<td>CAT ALS</td>
<td>(n = 15)</td>
<td>4 x weekly 1-1 ¼ hour sessions delivered at home by a trained practitioner in 4 themes: safe place, awareness, life chain, and perceptive. Self-hypnosis also encouraged.</td>
<td></td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Martin et al.</td>
<td>Mixed</td>
<td>CAT</td>
<td>(n = 25)</td>
<td>7 x 1 hour videoconference online CBT + self-hypnosis. Sessions delivered by a psychologist in groups of 4-5 participants every 2 weeks.</td>
<td></td>
<td></td>
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<td></td>
</tr>
</tbody>
</table>

**Life satisfaction and positive affect in gratitude condition, no differences for negative affect.**
<table>
<thead>
<tr>
<th>Aoun et al. 2015</th>
<th>Cohort</th>
<th>ALS Int: (n = 27)</th>
<th>Dignity Therapy</th>
<th>Delivered by a psychologist for patients &amp; caregivers at home.</th>
<th>Delivered in 3-7 visits (approx. 8 hours) over 14-113 days (Mdn = 36).</th>
<th>No control</th>
<th>HRQoL: ALSAQ-5.</th>
<th>No significant change in HRQoL or spiritual well-being from pre to 1 week post intervention.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Boosman et al. 2011</td>
<td>Cohort</td>
<td>Mixed Int: Total (n = 43)</td>
<td>Fatigue management group</td>
<td>Interactive small group educational sessions + homework on managing and coping with fatigue. Delivered by Occupational Therapist, Physiotherapist &amp; Social Worker at rehab clinic.</td>
<td>5 weekly 2 x hour sessions + 3 month follow-up session.</td>
<td>No control</td>
<td>HRQoL: SF-36</td>
<td>Significant improvements in SF-36 subscales role-physical, mental health and general health perceptions from baseline to 3 months post-intervention. Greater improvements in males and p’s with lower education levels &amp; lower baseline self-efficacy.</td>
</tr>
<tr>
<td>Palmieri et al. 2012 Cohort ALS Int: (n = 8)</td>
<td>Hypnosis-based intervention</td>
<td>Hypnosis-based intervention administered at home by a Psychologist (+ self-hypnosis). Related to issues such as illness acceptance &amp; resilience.</td>
<td>4 x weekly 45 min sessions (+ self hypnosis)</td>
<td>No control</td>
<td>HRQoL: ALSAQ-5, ALSSQOL-r</td>
<td>Significant improvements pre to immediately post intervention in ALSQOL-r total and subscales: religiosity and negative emotion. No significant change in ALSAQ-5.</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Abbreviations for conditions:** ALS = Amyotrophic Lateral Sclerosis; BD = Becker Dystrophy; CA = Cerebellar Ataxia; CMT = Charcot Marie Tooth Disease; ED = Emery-Dreifuss Dystrophy; FSHD = Facioscapulohumeral Dystrophy; HDM = Hereditary Distal Myopathy; HSP = Hereditary Spastic Paraparesis; KD = Kennedy Disease; LGD = Limb-Girdle Dystrophy; MD = Muscular Dystrophy; MND = Neuromuscular disorder; PD = Proximal Dystrophy; PPS = Post-Polio Syndrome; PN = Polynuropathy; SPA = Spinal Muscular Atrophy

**Abbreviations for measures:** ALSAQ-5; Amyotrophic Lateral Sclerosis Assessment Questionnaire-5 (Jenkinson et al., 2007); ALSAQ-40 = Emotional Functioning subscale of ALS Assessment Questionnaire (Jenkinson et al., 1999); ALSSQOL-r = Amyotrophic Lateral Sclerosis Specific Quality of Life – revised (Pagnini and Simmons, 2010); FACIT-sp 12 = Spiritual Well-being Scale of Functional Assessment of Chronic Illness Therapy (Peterman et al., 2002); HADS = The Hospital Anxiety and Depression Scale (Zigmond and Snaith, 1983); PWBQ = The Psychosocial Well-being Questionnaire (Kaasa et al., 1988); SF-36 = The Medical Outcome Study Short Form Health Survey (Ware and Sherbourne, 1992); SF-36 – MCS = Health Survey Short Form, Mental Component Summary; SIP = Sickness Impact Profile (Bergner et al., 1976); SWLS = Satisfaction with Life Scale (Diener et al., 1985); WHO-DAS II = World Health Organization Disability Assessment Schedule II (WHODAS Group, 2000)
Additional abbreviations: Con = Control group; CAT = Cohort Analytic Trial; CBT = Cognitive Behavioural Therapy; HRQoL = Health-related quality of life; Int = intervention group; M = Mean; n = number; p = participant; PWB = Psychological Well-being; QOL = Quality of life; RCT = Randomised Controlled Trial; SWB = Subjective Well-being; WB = Well-being
Quality Appraisal

Using the EPHPP Quality Assessment Tool for Quantitative Studies (Thomas et al., 2004), three RCTs were appraised as moderate in quality and the remainder were appraised as weak (see Table 2 for quality appraisal ratings). The methodological quality of studies was, therefore, generally inadequate.

Amongst the three moderate studies, small sample sizes, insufficient blinding, selective outcome reporting and use of a non-active control were problematic. In one study, a strong design was hampered by slow recruitment and retention resulting in the trial being stopped prematurely (Van Groentijn et al., 2015). Similar issues were identified in the seven studies appraised as weak. Of these studies, three used a non-equivalent control and three did not utilise any form of control. Most weak studies recruited participants from a single site or support group and thus are likely to be affected by selection bias. In many weak studies it was unclear whether co-intervention/contamination had occurred as this information was not clearly reported. Across all studies, blinding was particularly weak, with only two studies using blinded outcome assessors (Koopman et al., 2015; Van Groentijn et al., 2015). Overall, fidelity to the intervention was not adequately assessed. In contrast, data collection (e.g. use of validated scales etc.) was generally strong.
<table>
<thead>
<tr>
<th>Study</th>
<th>Selection Bias</th>
<th>Study Design</th>
<th>Confounds</th>
<th>Blinding</th>
<th>Data Collection</th>
<th>Withdrawals/ Dropouts</th>
<th>Global Rating</th>
<th>Additional Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ahlström et al. 2006</td>
<td>Moderate</td>
<td>Moderate</td>
<td>Weak</td>
<td>Weak</td>
<td>Strong</td>
<td>Strong</td>
<td>Weak</td>
<td>A small number of the control group were also receiving rehabilitation. No intervention protocol provided. Quality and consistency of intervention not measured.</td>
</tr>
<tr>
<td>Aoun et al. 2015</td>
<td>Weak</td>
<td>Moderate</td>
<td>N/A</td>
<td>Weak</td>
<td>Strong</td>
<td>Moderate</td>
<td>Weak</td>
<td>No control group. Detailed protocol for intervention provided and sessions recorded but not rated for quality and adherence.</td>
</tr>
<tr>
<td>Averill et al. 2013</td>
<td>Moderate</td>
<td>Strong</td>
<td>Strong</td>
<td>Weak</td>
<td>Moderate</td>
<td>Strong</td>
<td>Moderate</td>
<td>Selective outcome reporting. Standardised instructions provided but adherence to intervention not measured.</td>
</tr>
<tr>
<td>Study</td>
<td>Quality</td>
<td>Adherence</td>
<td>Intervention</td>
<td>Control</td>
<td>Quality</td>
<td>Adherence</td>
<td>Notes</td>
<td></td>
</tr>
<tr>
<td>-----------------------------</td>
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<td>------------------------------------------------------------------------------------------------</td>
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</tr>
<tr>
<td>Boosman et al. 2011</td>
<td>Weak</td>
<td>Moderate</td>
<td>N/A</td>
<td>Weak</td>
<td>Strong</td>
<td>Strong</td>
<td>Weak; No control group. Content of intervention described but no method of checking quality and adherence.</td>
<td></td>
</tr>
<tr>
<td>Emmons et al. 2003</td>
<td>Weak</td>
<td>Weak</td>
<td>Weak</td>
<td>Weak</td>
<td>Weak</td>
<td>Weak</td>
<td>Weak; Standardised instructions provided. Manipulation check confirmed gratitude condition elicited greater expressions of gratitude than control. Mean outcomes across intervention period calculated for analyses.</td>
<td></td>
</tr>
<tr>
<td>Kleinbub et al. 2015</td>
<td>Weak</td>
<td>Moderate</td>
<td>Strong</td>
<td>Weak</td>
<td>Strong</td>
<td>Strong</td>
<td>Weak; Small sample. Intervention protocol but no method of checking quality and adherence. 5 intervention participants were also receiving psychopharmacology</td>
<td></td>
</tr>
<tr>
<td>Koopman et al. 2015</td>
<td>Weak</td>
<td>Strong</td>
<td>Strong</td>
<td>Moderate</td>
<td>Strong</td>
<td>Moderate</td>
<td>Moderate; Small sample. Standardised CBT modules utilised and selected via criteria. Quality and adherence not assessed. Co-interventions received in control and treatment groups.</td>
<td></td>
</tr>
<tr>
<td>Study</td>
<td>Design Quality</td>
<td>Randomisation</td>
<td>Allocation Concealment</td>
<td>Blinding</td>
<td>Outcome Measurement</td>
<td>Outcome Reporting</td>
<td>Conclusion</td>
<td></td>
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<tr>
<td>Martínez et al. 2014</td>
<td>Weak</td>
<td>Moderate</td>
<td>Weak</td>
<td>Strong</td>
<td>Weak</td>
<td>Weak</td>
<td>Participants assigned to control group if they did not have access to a computer/webcam. Detailed protocol for intervention provided but no method of checking quality and adherence. Significant baseline differences in quality of life not controlled in analysis.</td>
<td></td>
</tr>
<tr>
<td>Palmieri et al. 2012</td>
<td>Weak</td>
<td>Weak</td>
<td>N/A</td>
<td>Weak</td>
<td>Moderate</td>
<td>Strong</td>
<td>Weak</td>
<td>No control group. Small sample size. Intervention protocol but no method of checking quality and adherence described.</td>
</tr>
<tr>
<td>Van Groenestijn et al. 2015</td>
<td>Weak</td>
<td>Strong</td>
<td>Strong</td>
<td>Moderate</td>
<td>Strong</td>
<td>Strong</td>
<td>Moderate</td>
<td>Small sample. Standardised CBT module utilised but no method of checking quality and adherence. Trial stopped prematurely due to slow recruitment and missing data.</td>
</tr>
</tbody>
</table>
**Study Findings: Effectiveness of Interventions**

Due to the heterogeneous nature of included studies, a meta-analysis was precluded. Instead, studies were grouped according to intervention type, outcome measurement and moderators of effectiveness as these groupings were deemed most pertinent to intervention effectiveness.

**Psychotherapy interventions.** Six studies investigated psychotherapy interventions, the most frequent of which was Cognitive Behavioural Therapy (CBT). One moderate quality RCT investigated CBT tailored to the management of fatigue in individuals with post-polio syndrome and self-reported severe fatigue (Koopman *et al.*, 2015). The study found no difference in HRQoL between participants randomised to CBT and a usual care control group immediately post-intervention and at the three and six month follow-up. The authors reported a slightly greater occurrence of co-interventions (e.g. medication, assistive devices etc.) in the control group, which could have potentially confounded the results. However, neither the CBT or control group showed any improvement over time. The authors suggest that as post-polio syndrome has an early onset, acceptance of fatigue may develop over time, thus reducing the effectiveness of intervention (Koopman *et al.*, 2015).

In contrast, another moderate quality RCT investigated CBT based on a stress-coping model for patients with ALS and their partners (Van Groenestijn *et al.*, 2015). The study found that, compared to a non-active control, intervention patients’ mental quality of life deteriorated less over time between baseline and six month follow-up. However, this effect was only found in one of the two measures used. It should also be
noted that the very small sample size in this study (total $n = 15$) means that the statistical tests utilised were not adequately powered (Van Groenestijn et al., 2015). The contribution of these findings to the evidence base is therefore questionable.

Additionally, a CAT appraised as weak in quality investigated the efficacy of a CBT-based online intervention delivered via video conference (Martínez et al., 2014). The intervention was tailored to neuromuscular disorders and involved psychoeducation, managing emotional reactions, cognitive restructuring, problem solving and relaxation exercises for groups of patients with mixed neuromuscular disorders. Across the three different measures of HRQoL, the authors reported the intervention group significantly improved between baseline and follow-up (4 ½ months later) on overall HRQoL and most subscales. It should be noted, however, that the intervention group had significantly lower levels of baseline HRQoL than the control group and at follow-up the two groups were comparable. As appropriate statistical analyses were not conducted to calculate the group X time interaction or control for baseline differences, the results may simply reflect regression to the mean in the intervention group.

Two linked, weak quality studies investigated the effect of a hypnosis intervention on quality of life and psychological well-being in patients with ALS. The first was a small pilot cohort study which assessed the impact of four sessions of Psychologist delivered hypnosis around issues such as illness acceptance, resilience and physical symptomology control, in addition to self-hypnosis training (Palmieri et al.,
In the absence of a control group, the authors reported significant improvements in one measure of HRQoL and subscales negative emotion and religiousness/spirituality. However, only eight participants took part in the study and therefore statistical tests were not adequately powered. Moreover, effects were not replicated in the second measure of HRQoL utilised in the study. Generalisability of the findings is therefore limited. This intervention was subsequently extended to a brief psychodynamic-based hypnosis intervention with four sessions on topics labelled safe place, awareness, life chain and perceptive (Kleinbub et al., 2015). Assessing the impact on HRQoL for 15 intervention participants and 15 matched controls, the authors reported a small effect of treatment on HRQoL immediately post intervention and at three months, with scores reverting towards baseline levels at six months. It should be noted however, that one third of the intervention group participants were receiving a low dose antidepressant or anxiolytic which could potentially confound the results.

The final weak cohort study assessed the impact of dignity therapy on HRQoL and spiritual well-being in patients with ALS and their caregivers (Aoun et al., 2015). Dignity therapy involves an in-depth interview with patients on their accomplishments in life and things they would like to pass on to loved ones which is later transcribed to create a permanent record for the individual and their family (Aoun et al., 2015). In a pre-post design the authors reported no significant impact on HRQoL or spiritual well-being one week post-intervention. Results are difficult to interpret due to the absence of a control. Moreover, the absence of an effect could potentially be influenced by the
short follow-up time and brief measure used to assess HRQoL which may not be
sensitive to change.

In summary, evidence for the effectiveness of psychotherapy interventions was
mixed, with some types of psychotherapy found to be beneficial for quality of life and
well-being (e.g. CBT and hypnosis) and some types found to be ineffective (e.g. CBT for
fatigue and dignity therapy). Study quality was generally higher for CBT-related
interventions than other forms of psychotherapy, however all studies had considerable
methodological limitations.

Writing interventions. Two studies examined the effects of brief writing
interventions on well-being. One moderate quality RCT investigated expressive
disclosure in patients with ALS (Averill et al., 2013). Participants randomised to the
intervention either wrote or audio recorded themselves speaking about their deepest
thoughts in relation to their condition for three days in a one week period. Control
participants completed outcome measures only. The authors reported a significant
increase in psychological well-being from pre-post intervention in the expressive
disclosure group, which was maintained at three months. In contrast, the control group
experienced deterioration in psychological well-being from pre-post intervention which
was also maintained at three months. Both groups reverted to near baseline levels at
six months suggesting the effectiveness of the intervention was not maintained beyond
three months. Furthermore, the use of a non-active control and selective outcome
reporting (individual measures used to create a composite ‘psychological wellbeing’ score are not reported) limit the generalisability of findings.

A writing intervention with a different emphasis was utilised by one CAT appraised as weak in quality (Emmons et al., 2003). In this gratitude intervention, participants with slowly progressive neuromuscular conditions (Charcot-Marie-Tooth disease, post-polio syndrome & facioscapulohumeral dystrophy) were asked to write down five things they were grateful for and complete experience ratings daily for 21 days. Control participants completed the experience ratings but were not asked to write gratitude lists. The study found the intervention group had significantly higher subjective well-being than the control group across the 21 day study period. It should be noted, however, that a three-item measure was developed by the author to assess subjective well-being and the reliability and validity of this measure are unknown. This study also assessed observer reports of well-being, with caregivers/ spouses asked to complete measures of life satisfaction and positive and negative affect from the patient’s perspective at the end of the intervention. The authors reported that life satisfaction and positive affect were rated significantly higher for intervention group patients compared with ratings for patients in the control group; no differences were reported for negative affect. The absence of any follow-up measures for this intervention beyond the 21 day study period mean it is not possible to establish whether effects were maintained.
In summary, there was some evidence for the effectiveness of expressive writing and gratitude lists in improving well-being, albeit in the short-term. As with psychotherapy interventions, the quality of evidence for the effectiveness of writing interventions was limited.

**Group psychoeducation/comprehensive rehabilitation.** Two weak quality studies assessed group-based psychosocial interventions delivered in clinic settings. One cohort study assessed an educational fatigue management intervention for patients with mixed slowly progressive neuromuscular conditions or multiple sclerosis (Boosman *et al.*, 2011). Between baseline and three months post intervention significant improvements were found in role-physical, mental health and general health perceptions subscales of the HRQoL measure SF-36 (Ware and Sherbourne, 1992). The absence of a control group makes the results difficult to interpret. The second study did utilise a control group (usual care) and found no significant change in HRQoL, psychological well-being or mood following a 10 day comprehensive rehabilitation programme for adults with muscular dystrophy (Ahlström *et al.*, 2006). It should be noted that a small number of the control group were also receiving rehabilitation which was not controlled for in the analyses.

In summary, whilst there was weak evidence for the effectiveness of group-based psychoeducation and the ineffectiveness of comprehensive rehabilitation, the absence of appropriate controls within studies inhibited meaningful interpretation of findings.
Study Findings: Assessment of Quality of Life and Well-Being

Quality of life and well-being were defined and assessed differently across all included studies, thereby complicating interpretation of findings (see Table 1). In one study, psychological well-being was considered a multi-faceted concept incorporating HRQoL (Averill et al., 2013). Conversely, in another study, quality of life was defined and assessed as a multi-faceted concept incorporating psychological well-being (Ahlström et al., 2006). The majority of studies on ALS utilised one of several ALS specific measures of HRQoL. Both studies utilising the ALSAQ-5 (Jenkinson et al., 2007) reported no significant effects, despite effects found in other similar measures in one of these studies (Aoun et al., 2015, Palmieri et al., 2015). This suggests that different measures of HRQoL may be measuring different constructs. Amongst other neuromuscular conditions, the SF-36 (Ware and Sherbourne, 1992), and SIP (Bergner et al., 1976) were most commonly used to assess generic HRQoL. The two studies utilising a specific validated measure of well-being (as opposed to a composite measure or author developed scale) did not report any beneficial effects (Aoun et al., 2015; Ahlström et al., 2006). Overall, outcome measurement was problematic and lacked a consistent approach across studies.
Study Findings: Moderators of Effectiveness

Only two studies investigated factors which may have moderated effectiveness of the intervention on quality of life and/or well-being. Averill et al. (2013) reported that higher ambivalence over expressing emotion was associated with greater increases in psychological-wellbeing three months after expressive disclosure. Boosman et al. (2011) reported greater improvements in HRQoL following a fatigue management intervention in participants who were male, individuals with lower education levels and those with lower baseline self-efficacy.

Components of each intervention were extracted and compared to allow for the identification of possible moderators of effectiveness (see Table 3). No clear pattern emerged from this analysis. Further research examining potential moderating factors is, therefore, required.
### Table 3: A comparison of intervention factors within included studies

<table>
<thead>
<tr>
<th>Study</th>
<th>Theoretical basis specified</th>
<th>Protocol/Manual</th>
<th>Delivered by Psychologist</th>
<th>Provides psycho-education</th>
<th>Targets emotional expression</th>
<th>Targets relaxation</th>
<th>Targets cognitions</th>
<th>Targets behaviours</th>
<th>Facilitates social support</th>
</tr>
</thead>
<tbody>
<tr>
<td>Koopman et al. 2015</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td></td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Averill et al. 2013</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Van Groenestijn et al. 2015</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>X</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Ahlstöm et al. 2006</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Emmons et al. 2003</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Kleinbub et al. 2015</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Martinez et al. 2014</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Aoun et al. 2015</td>
<td>X</td>
<td>✓</td>
<td>✓</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Boosman et al. 2011</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Palmieri et al. 2012</td>
<td>X</td>
<td>✓</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>✓</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
</tbody>
</table>

= No significant effects of intervention on quality of life and/or well-being
Discussion

Key Findings

This systematic review sought to synthesise the available evidence for the effectiveness of psychosocial interventions on quality of life and wellbeing in adults with neuromuscular disorders. Furthermore, it sought to critically appraise the methodological quality of this evidence. Despite calls for research on psychosocial interventions in this patient group (Graham et al., 2015; Pagnini et al., 2012), only a small number of studies have been conducted. Of the 10 included studies, seven reported a beneficial effect of psychosocial interventions across different neuromuscular disorders. Overall, there is mixed and contradictory evidence for the effectiveness of CBT in improving HRQoL and mental quality of life, with moderate evidence of its ineffectiveness in severely fatigued participants (Koopman et al., 2015; Van Groenestijn et al., 2015; Martínez et al., 2014;). It should be noted that CBT is the only intervention where a beneficial effect was found to be maintained at six months post intervention (Van Groenestijn et al., 2015); however, this may reflect a paucity of longer-term follow-up data in other studies. In the short-term, there was moderate evidence for emotional disclosure improving psychological well-being (Averill et al., 2013), weak evidence for gratitude lists improving subjective well-being (Emmons et al., 2003), and weak evidence for hypnosis (Kleinbub et al., 2015; Palmieri et al., 2012) and a fatigue psychoeducational group (Boosman et al., 2011) improving HRQoL. In contrast, there was weak evidence to suggest that dignity therapy does not improve
HRQoL and spiritual well-being (Aoun et al., 2015) and that comprehensive rehabilitation does not improve global quality of life (Ahlstöm et al., 2006). In summary, there is currently insufficient evidence to support the use of psychosocial interventions to improve quality of life and well-being in adults with neuromuscular disorders.

The majority of included studies were rated as weak in quality, with only three rated as moderate and none as strong. Across studies, factors relating to selection bias, study design and blinding were particularly problematic. The majority of studies recruited a small number of participants, often from a single source. Whilst this may reflect the rarity of neuromuscular conditions and the challenges they pose to engagement in activities (Hilton-Jones, Freebody and Stein, 2011), the risk of biased sampling limits generalisability of findings. With respect to study design, no included studies utilised an active control and three studies included no form of control. Thus, positive findings could be due to non-specific treatment effects. Importantly, as many studies did not assess long-term outcomes of interventions, inferences regarding long-term effects cannot be made. In most studies there was no blinding, with participants and outcome assessors fully aware of the research question and group allocations. Furthermore, consistency of the intervention was rarely assessed beyond the development of treatment protocols. In the context of these numerous risks of bias, caution must be exercised in interpretation of findings.
Further challenges to useful synthesis within this systematic review arose from the widespread heterogeneity amongst included studies. In particular, contrasting definitions and assessment methods for quality of life and well-being impeded meaningful comparison across studies. It is possible that this is reflective of confusion surrounding these terms within the wider literature and the field of neuromuscular disease (Burns et al., 2012; Camfield and Skevington, 2008). HRQoL was the most commonly assessed outcome, however many different measures were utilised. Across all outcomes, a diverse group of measures were employed including both disorder specific and generalised measures and both single measures and multiple/composite measures. Of further concern, some studies reported contradictory findings when different measures were utilised to assess the same construct within an individual study (e.g. Van Groenestijn et al., 2015; Palmieri et al., 2012), thereby providing evidence against the comparability of outcome measures. Thus, the extent to which quality of life and well-being were validly and reliably assessed is cause for concern.

Identifying potential moderators of outcomes is an important aspect of intervention research. However, findings relevant to moderator variables were limited. Moderate evidence suggests that greater benefits to psychological well-being were gained by individuals with ambivalence over emotional expression (Averill et al., 2013). Additionally, limited evidence suggests that greater benefits to HRQoL were gained by men, individuals with lower levels of education, and those with poor self-efficacy (Boosman et al., 2011). At the intervention level, no component emerged as consistently associated with gains in quality of life or well-being. It is, therefore, not
possible to conclude whether particular characteristics of participants and/or interventions are most likely to result in beneficial effects from psychosocial interventions.

**Implications for Research**

Whilst still at an early stage, research in this field has developed rapidly in recent years, with all three randomised controlled trials published in the last two years. Although these developments are promising, methodological limitations are still endemic in the available literature. It is vital that research proceeds with a strong emphasis on study quality such that meaningful conclusions can be made. As the gold standard for intervention research, randomised controlled trials should be prioritised, with double-blind designs and multi-site recruitment employed where possible. Active controls should also be utilised to establish whether beneficial effects are due to the specific intervention or simply from non-specific effects associated with being part of an intervention. In developing interventions, researchers should ensure detailed protocols are utilised and treatment fidelity is assessed.

Given the degenerative nature of neuromuscular diseases (Hilton-Jones, Freebody and Stein, 2011), it is vital that long term outcomes of interventions are assessed. Additionally, to avoid compounding current confusion, researchers should clearly define outcomes of interest according to accepted understandings of quality of life and well-being. Validated, condition-specific measures of quality of life may be preferable to generalised measures (Burns et al., 2012). Moreover, since HRQoL is
currently the most frequently assessed outcome, greater emphasis on well-being would be useful to progress understanding of the potential impact of interventions in this patient population.

To further progress understanding, future research could also usefully include analysis of moderator variables. For example, researchers could investigate which individuals are most likely to benefit from an intervention by assessing the impact of individual difference and demographic factors on outcomes.

**Implications for Clinical Practice and Policy**

There is currently insufficient evidence to conclusively recommend psychosocial interventions for adults with neuromuscular disorders. However, since no study reported adverse outcomes from intervention, it can be concluded that clinicians and researchers should proceed in the development and evaluation of appropriately designed psychosocial interventions. Whilst one of the three moderate quality studies reported no beneficial effect of CBT, it should be noted that this intervention focused only on management of fatigue, which the authors acknowledged was not an important goal for all participants (Koopman et al., 2015). Given the individual variation and broad impact of neuromuscular disorders in adulthood (Hilton-Jones, Freebody and Stein, 2011), it is imperative that psychosocial interventions are designed and delivered in a way that is patient-centred.

**Strengths and Weaknesses of this Review**
This review extends findings from the one previous review in this area (Gould et al., 2015) to synthesise current knowledge on the impact of psychosocial interventions on quality of life and well-being in adults with neuromuscular disorders. The broad and systematic search strategy is a particular strength of this review. The main limitation of the current review is the heterogeneity amongst included studies. Due to the various included study designs and outcomes measures, a meta-analysis could not be performed. Consequently, no statistical inferences regarding effectiveness can be made.

Conclusion

There is currently no strong evidence to determine whether psychosocial interventions improve quality of life and well-being in adults with neuromuscular disorders. Although some benefits to both quality of life and well-being have been identified from a number of psychosocial interventions, such benefits are almost exclusively short-term and subject to bias. The paucity of high quality research and confusion surrounding measurement of quality of life and well-being impede meaningful interpretation of findings. Further research with a strong emphasis on study quality, clearly defined outcomes and consideration of moderator variables is urgently required to progress current understanding and inform recommendations for practice. Given that the overall prevalence of neuromuscular disorders in adulthood is increasing (Deenen et al., 2015), it is vital that the identification of effective, patient-centred interventions becomes a prominent research priority.
References


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doi:10.1016/S0738-3991(02)00213-6


doi:10.1177/0269215513487234

Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration. 16(5-60), pp. 1–7. doi:10.3109/21678421.2015.1038276


Systematic Review: Appendix A - Full search strategy
PsycInfo search strategy (yielded 816 results):

PsycInfo was searched in June 2015 using the following search strategy:

1. ("neuromuscular disorder*" OR "neuromuscular*" OR "muscular dystrophy" OR "myasthenia Gravis" OR "charcot-marie-tooth" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in any field

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR counseling OR "psychological therapy" OR psychotherapy OR psychodynamic OR "cognitive behavior*" OR cognitive OR behavior* OR CBT OR "self management" OR "self care" OR mindfulness OR "acceptance and commitment therapy" OR tele*): in any field

3. AND ("quality of life" OR "well being" OR "well-being" OR "wellbeing" OR "life satisfaction" OR "mental health" OR "psychological stress" OR anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "psychological adjustment"): in any field

Medline search strategy (yielded 1859 results):

Medline was searched in June 2015 using the following search strategy:

1. ("neuromuscular disorder*" OR "neuromuscular*" OR "muscular dystrophy" OR "myasthenia Gravis" OR "charcot-marie-tooth" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in any field

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR counseling OR "psychological therapy" OR psychotherapy OR psychodynamic OR "cognitive behavior*" OR cognitive OR behavior* OR CBT OR "self management" OR "self care" OR mindfulness OR "acceptance and commitment therapy" OR tele*): in any field

3. AND ("quality of life" OR "well being" OR "well-being" OR "wellbeing" OR "life satisfaction" OR "mental health" OR "psychological stress" OR anxiety OR...
depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "psychological adjustment"): in any field

PsycArticles search strategy (yielded 21 results):

PsycArticles was searched in June 2015 using the following search strategy:

1. ("neuromuscular disorder*" OR "neuromuscular*" OR "muscular dystrophy" OR "myasthenia Gravis" OR "charcot-marie-tooth" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in any field

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR counseling OR "psychological therap*" OR psychotherapy OR psychodynamic OR "cognitive behavio*r*" OR cognitive OR behavio*r* OR CBT OR "self manag*" OR "self care" OR mindfulness OR "acceptance and commitment therapy" OR tele*): in any field

3. AND ("quality of life" OR "well being" OR "well-being" OR "wellbeing" OR "life satisfaction" OR "mental health" OR "psychological stress" OR anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "psychological adjustment"): in any field

CINAHL search strategy (yielded 704 results):

CINAHL was searched in June 2015 using the following search strategy:

1. ("neuromuscular disorder*" OR "neuromuscular*" OR "muscular dystrophy" OR "myasthenia Gravis" OR "charcot-marie-tooth" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in any field

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR counseling OR "psychological therap*" OR psychotherapy OR psychodynamic OR "cognitive behavio*r*" OR cognitive OR behavio*r* OR CBT OR "self manag*" OR "self care" OR mindfulness OR "acceptance and commitment therapy" OR tele*): in any field
3. AND ("quality of life" OR "well being" OR "well-being" OR "wellbeing" OR "life satisfaction" OR "mental health" OR "psychological stress" OR anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "psychological adjustment"): in any field

**AMED search strategy** (yielded 130 results):

AMED was searched in June 2015 using the following search strategy:

1. ("neuromuscular disorder*" OR "neuromuscular*" OR "muscular dystrophy" OR "myasthenia Gravis" OR "charcot-marie-tooth" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in any field

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR counsel#ing OR "psychological therap*" OR psychotherapy OR psychodynamic OR "cognitive behavio$r*" OR cognitive OR behavio$r* OR CBT OR "self manag*" OR "self care" OR mindfulness OR "acceptance and commitment therapy" OR tele*): in any field

3. AND ("quality of life" OR "well being" OR "well-being" OR "wellbeing" OR "life satisfaction" OR "mental health" OR "psychological stress" OR anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "psychological adjustment"): in any field

**Web of Science search strategy** (yielded 344 results):

Web of Science was searched in June 2015 using the following search strategy:

1. ("Neuromuscular Disorder*" OR Neuromuscular* OR "Muscular Dystrophy" OR "Myasthenia Gravis" OR "CHARCOT-MARIE-TOOTH" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in topic and title

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR Counse$l*ing OR "psychological therap*" OR Psychotherapy OR psychodynamic OR Cognitive Behavio$r* OR Cognitive OR Behavio$r* OR CBT OR "Self Manag*" OR "self
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care" OR mindfulness OR "Acceptance and Commitment Therapy" OR Tele*): in topic and title

3. AND ("Well Being" OR "Well-being" OR "Wellbeing" OR "Life Satisfaction" OR "Mental Health" OR "Quality of Life" OR "psychological stress" OR Anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "Psychological adjustment"): in topic and title

4. AND Database excluded: Medline

The Cochrane Register of Controlled Trials (CENTRAL) search strategy (yielded 81 results):

The Cochrane Register of Controlled Trials was searched in June 2015 using the following search strategy:

1. ("Neuromuscular Disorder*" OR "Neuromuscular*" OR "Muscular Dystrophy" OR "Myasthenia Gravis" OR "CHARCOT-MARIE-TOOTH" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome"): in title, abstract and keywords

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR Counsel*ing OR "psychological therap*" OR Psychotherapy OR psychodynamic OR "Cognitive Behavio*r*" OR Cognitive OR Behavio*r OR CBT OR "Self Manag*" OR "self care" OR mindfulness OR "Acceptance and Commitment Therapy" OR Tele*): in title, abstract and keywords

3. AND ("Well Being" OR "Well-being" OR Wellbeing OR "Life Satisfaction" OR "Mental Health" OR "Quality of Life" OR "psychological stress" OR Anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "Psychological adjustment"): in title, abstract and keywords

Open Grey Search strategy (yielded 15 results):

Open Grey was searched in June 2015 using the following search strategy:

1. ("Neuromuscular Disorder*" OR "Neuromuscular*" OR "Muscular Dystrophy" OR "Myasthenia Gravis" OR "CHARCOT-MARIE-TOOTH" OR
"amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome")

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR "Counseling" OR "counselling" OR "psychological therap*" OR Psychotherapy OR psychodynamic OR "Cognitive Behavior*" OR "Cognitive Behaviour*" OR "Cognitive" OR "Behavior*" OR "Behaviour*" OR CBT OR "Self Manag*" OR "self care" OR mindfulness OR "Acceptance and Commitment Therapy" OR Tele*)

3. AND ("Well Being" OR "Well-being" OR "Wellbeing" OR "Life Satisfaction" OR "Mental Health" OR "Quality of Life" OR "psychological stress" OR Anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "Psychological adjustment")

**Conference Abstracts Index** search strategy (yielded 160 results):

Conference Abstracts Index was searched in June 2015 using the following search strategy:

1. (Neuromuscular Disorder* OR "Neuromuscular*" OR "Muscular Dystrophy" OR "Myasthenia Gravis" OR "CHARCOT-MARIE-TOOTH" OR "amyotrophic lateral sclerosis" OR "motor neuron disease" OR "spinal muscular atrophy" OR "post polio syndrome")

2. AND (psychosocial OR psychological OR social OR "Social Support" OR "Support Group*" OR psychoeducation OR "peer support" OR "Counseling" OR counselling OR "psychological therap*" OR Psychotherapy OR psychodynamic OR "Cognitive Behaviour*" OR "cognitive behavior*" OR "Cognitive" OR "Behaviour*" OR "Behaviour*" OR CBT OR "Self Manag*" OR "self care" OR mindfulness OR "Acceptance and Commitment Therapy" OR Tele*)

3. AND (Well Being OR "Well-being" OR "Wellbeing" OR "Life Satisfaction" OR "Mental Health" OR "Quality of Life" OR "psychological stress" OR Anxiety OR depression OR pain OR fatigue OR "social participation" OR "positive psychology" OR "Psychological adjustment")
## Systematic Review: Appendix B - Data extraction form

<table>
<thead>
<tr>
<th>Article</th>
<th>Study Design</th>
<th>Participants</th>
<th>Study Design</th>
<th>Quality tool screening score</th>
<th>Total number and response rate</th>
<th>Age</th>
<th>Sex</th>
<th>NMD diagnoses (type, duration)</th>
<th>Inclusion/exclusion criteria</th>
<th>Countries</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Interventions</th>
<th>Measures</th>
<th>Outcomes</th>
<th>Miscellaneous</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type of intervention</td>
<td>Delivery of Intervention (format/how delivered/by whom/where)</td>
<td>Comparison</td>
<td>Participants and drop outs per intervention</td>
</tr>
</tbody>
</table>
Systematic Review: Appendix C - Flow diagram of study selection process

Figure 1: Flow diagram of study selection process

Records identified through database searching + email alerts (n = 4130)

Additional records identified through other sources (e.g. contacting experts, reference lists etc) (n = 5)

Records after duplicates removed (n = 3136)

Records excluded as clearly irrelevant (n = 3,117)

Records screened (n = 3136)

Full-text articles assessed for eligibility (n = 19)

Full-text articles excluded: (n = 9)
  - Intervention not psychosocial (4)
  - Quality of life/well-being not assessed (2)
  - Duplication of data (1)
  - No results included (1)
  - Majority of participants did not have a NMD (1)

Studies included in narrative synthesis (n = 10)
Appendix B – Ethical approval documentation

I Ethical approval letter

UWE REC REF No: HAS.16.10.034

11th November 2016

Elaine Walklet
Psychology Department
St John’s Campus
Henwick Grove
University of Worcester
Worcs  WR2 6AJ

Dear Elaine

Application title: Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: a Photovoice study

Your ethics application was considered by the Faculty Research Ethics Committee and, based on the information provided, has been given ethical approval to proceed.

The scrutineer noted that on the online consent form there is an issue of formatting where the check boxes obscure some of the text.

You must notify the committee in advance if you wish to make any significant amendments to the original application using the amendment form at http://www1.uwe.ac.uk/research/researchethics/applyingforapproval.aspx
Please note that any information sheets and consent forms should have the UWE logo. Further guidance is available on the web: [http://www1.uwe.ac.uk/aboutus/departmentsservices/professionalservices/marketingandcommunications/resources.aspx](http://www1.uwe.ac.uk/aboutus/departmentsservices/professionalservices/marketingandcommunications/resources.aspx)

The following standard conditions also apply to all research given ethical approval by a UWE Research Ethics Committee:

1. You must notify the relevant UWE Research Ethics Committee in advance if you wish to make significant amendments to the original application: these include any changes to the study protocol which have an ethical dimension. Please note that any changes approved by an external research ethics committee must also be communicated to the relevant UWE committee.

2. You must notify the University Research Ethics Committee if you terminate your research before completion;

3. You must notify the University Research Ethics Committee if there are any serious events or developments in the research that have an ethical dimension.

Please note: The UREC is required to monitor and audit the ethical conduct of research involving human participants, data and tissue conducted by academic staff, students and researchers. Your project may be selected for audit from the research projects submitted to and approved by the UREC and its committees.

We wish you well with your research.

Yours sincerely

Dr Julie Woodley
Chair
Faculty Research Ethics Committee

c.c. Tim Moss
II Participant Information Sheet

Participant information sheet

Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: a Photovoice study

We would like to invite you to take part in our research study. Before you decide whether to take part we would like you to understand why the research is being done and what it would involve for you. Please feel free to contact the lead researcher if you have any questions about the study:

Name: Elaine Walklet

Telephone number: 01905 542420

Email: e.walklet@worc.ac.uk

What is this study about?

Research suggests that living with Charcot-Marie Tooth (CMT) disease can affect an individual’s quality of life. ‘Quality of life’ is difficult to define however, and is also difficult to measure. In order to improve our understanding and assessment of quality of life in adults with CMT, it is important that we gather in-depth accounts from people who live with the condition. This study aims to use a new research method called ‘Photovoice’ to identify what adults with CMT think about their quality of life and factors that influence this (positively or negatively) over time. In order to achieve this, this study involves taking digital photographs (using a smart phone or digital camera) and writing about them in a secure, online journal over a 2 week period.

Why have I been invited to take part?

You have been invited to take part because you are over 18 years old and have a diagnosis of CMT (any type). You are one of a small number of people that will be invited to take part.

Do I have to take part?

No. It is entirely up to you to decide whether you would like to join the study. Please take your time to decide if you would like to take part. If you agree to participate, we will then ask you to
sign a consent form. You are free to later withdraw from the study, without giving a reason. This will not affect any care or treatment you may be receiving.

**What will happen to me if I take part?**

You will be asked to complete a consent form and provide some basic details about yourself, including your email address. You will then be contacted with information and log-in details to access a secure and anonymous online journal page, accessible only by you, the lead researcher and the University of Worcester learning technology team who have signed a data processing agreement to set up your account. You will be asked to use the journal to upload digital photographs of anything which you feel affects your quality of life (positively, negatively or neutral) during a 2 week period. It is up to you how frequently you post to the website and how many photographs you upload in total, although we would suggest a maximum of 25. Importantly, we would like you to write a diary entry in the journal for each photograph you upload, explaining what the photograph represents, why it is important to you and how it influences your quality of life. We will let you know that your journal entries are being received at the start, middle and end of the 2 weeks. After the 2 week period you will also receive an email thanking you for your participation.

**Expenses and payments**

We do not anticipate that taking part in the study will cost you anything. As this is not a funded research project, unfortunately we are not able to provide payment for your involvement in this study.

**What are the possible benefits of taking part?**
We cannot promise the study will help you, but the information we get from this study may help to better understand quality of life for people with CMT and factors which influence it. In the long term, we hope this will improve management of the condition.

**What are the possible disadvantages of taking part?**
The main disadvantage is that the study will take some time to complete. Each upload of a photograph and supporting diary entry is likely to take approximately 5 minutes. Another potential disadvantage is that thinking about factors which negatively affect you quality of life could cause you distress. Sources of support are provided below should you feel in any way distressed by the experience.

Samaritans: http://www.samaritans.org (08457 90 90 90) – Available 24 hours/ day, 7 days/week

MIND: www.mind.org.uk (0300 123 3393) – Available Monday-Friday, 9am-6pm

**Will my information be kept confidential?**
All information which is collected about you during the course of the research will be handled in strict confidence. Only the lead researcher will have access to your full personal information. Your name and email address (but no other personal details) will be securely transferred to the journal administrator who will set up your account. Only the lead researcher and journal administrators will have access to your journal. Your anonymised photographs and diary entries will be securely downloaded and stored with a unique code to protect your identity. They will then be reviewed within the research team as part of the analysis for the study. Within publications of the study, direct quotes and photographs may be used to support explanations of any themes which emerge. In this instance you will not be referred to by your name or any other identifiable information. After 5 years, all data will be destroyed.

The only exception to this confidentiality agreement would be if the research team were concerned about your safety. If you disclose something which suggests that you or someone else may be at risk of harm we will have to take appropriate advice. In this instance the research team may need to speak to a medical professional but would always discuss this with you first.
What will happen to the results of the study?

The information you provide will be analysed and then combined with information provided by other participants. The study will be written up for publication and conference presentation. You will not be identified in any of these documents.

What will happen if I don’t want to carry on with the study?

If you decide you no longer wish to take part, you have the right to withdraw your data in the 4 weeks after the study period has ended. In this instance, please contact lead researcher Elaine Walklet (email: e.walklet@worc.ac.uk / tel: 01905 542420) with your participant log-in and request your data be withdrawn; you will not need to give a reason and there will not be any consequences. Any withdrawn data will be destroyed and will not contribute to the final analysis.

What if there is a problem?

If you have a concern about any aspect of this study, you should ask to speak to the researchers who will do their best to answer your questions [email: e.walklet@worc.ac.uk / tel: 01905 542420]. If you remain unhappy and wish to complain formally, you can contact:

Dr Tim Moss, Director of Postgraduate Research in Health and Social Sciences, University of the West of England, Frenchay Campus Coldharbour Lane Bristol BS16 1QY

Email: Tim.Moss@uwe.ac.uk

Tel: +44 (0)117 32 82189

Who is organising and funding this research?

This research is organised by the University of the West of England. This research is not externally funded.

Who has reviewed the study?

This study has been reviewed and given a favourable opinion by the Health and Social Sciences Faculty Research Ethics Committee.
Further information and contact details

If you would like specific information about this research project, please contact:

Elaine Walklet, Lecturer in Health Psychology, University of Worcester, Henwick Grove, Worcester, WR2 6AJ.

Email: e.walklet@worc.ac.uk

Tel: 01905 542420
Study: Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: a Photovoice study

1. I confirm that I have read and understand the information sheet for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.

2. I understand that my participation is voluntary and that I am free to withdraw up to 4 weeks after completing the study without giving any reason and without my medical care or legal rights being affected.

3. I understand that if I disclose something which indicates an intention to harm myself or others that the research team will seek medical advice following discussion with me.

4. I understand that I will not be made identifiable in reports of the study, including publication of the study and its findings.

5. I am 18 years of age or older and do not have a diagnosis of any of the following conditions which could affect my capacity to consent: dementia, brain damage, severe learning disability or severe mental health condition such as schizophrenia or bipolar disorder.

6. I agree that direct (anonymised) quotes from my diary entries can be used in reports of the study.

7. I agree to ask full verbal permission before taking photographs of other adults in places where privacy could be reasonably expected. I understand that photographs of identifiable adults will not be published in the write-up of this study but will be used in the analysis.

8. I agree to ask full verbal permission from parents/guardians before taking photographs of children. I understand that photographs of identifiable
children will not be published in the write-up of this study but will be used in the analysis.

9. I grant the research team full permission to use and publish my photographs within the analysis and dissemination of the study (e.g. doctorate thesis, journal articles, book chapters, conference presentations etc.). I understand that my photographs will only be used for the purposes described in the participant information sheet.

10. I give my permission for my name and email address to be securely transferred to a technologist at the University of Worcester who will set up my online blog account.

11. I agree to take part in the above study.

12. I consent to be contacted by the research team after I have completed the study.

IV Online Participant Details Form

ONLINE PARTICIPANT DETAILS FORM

Thank you for consenting to take part in our study. Please complete the following details to register for the study:
1. Title: _______

2. Full Name: ______________________________

3. Email address for project registration: ________________

4. Gender: Male/Female/Transgender/ Prefer not to say

5. Date of Birth (XX/XX/XX): __________

6. Nationality: _______________________

7. Occupational status: Employed/self-employed full-time/ Employed self-employed part-time/ Unemployed/ Homemaker/ Retired/ Other or prefer not to say

8. Type of CMT: 1A/1B/1C/1D/1F/1X/2A/2B/2C/2D/2E/3/4A/4B/4C/4D/4F/Other/ Unknown

9. Age at diagnosis: ______

10. Do other members of your family have CMT? Yes/No/Unsure

11. Please create a pseudonym (a fictional name which will be used in the write up of the study to protect your identity)________________________________________

Thank you for completing these details. A member of the research team will contact you by email shortly with your log-in details for the online journal and information about how to use it. If you do not receive an email within a few days, please check your junk mail and contact lead researcher Elaine Walklet (e.walklet@worc.ac.uk).
V Information about photovoice

Information about Photovoice

Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: a Photovoice study

Photovoice is a relatively new research method which involves photographing aspects of experiences over a short period of time and then writing about these or talking about them with a researcher. Research into other chronic health conditions has found that this can provide new insights into important factors that other research methods can overlook.

As part of this study, we would like you to take digital photographs (e.g. using a smart phone or digital camera) of anything which you feel affects your quality of life (positively, negatively or neutral) during a 2 week period. It is up to you how frequently you post to the website/app and how many photographs you upload in total, although we would suggest a maximum of 25. Importantly, we would like you to write a diary entry for each photograph you upload explaining what the photograph represents, why it is important to you and how it influences your quality of life. Once you have signed your consent form, we will send you log-in details to a secure online journal so that you can begin to take and upload your photos and diary entry. We will provide full instructions on how to do this and contact details if you have any questions about using the journal.

Taking photographs – important information!

As you may be aware, there are laws and regulations related to taking photographs. Before you begin, please ensure you familiarise yourself with the following information:

- Please be mindful of your personal safety and security when taking photographs and in the event you ask permission from someone to photograph them.

- Please ensure you ask full verbal permission before taking photographs of adults in areas where privacy could be reasonably expected (e.g. in their home or garden).
• Adults should read your information sheet and are welcome to contact the lead researcher for more information (Elaine Walklet: e.walklet@worc.ac.uk/ 01905 542420). Please note that taking photographs of adults in public areas where privacy could not be reasonably expected does not require such consent. Photographs of identifiable adults will not be published in the write-up of this study but will be used within the analysis.

• Please ensure you always ask full verbal permission from a parent/guardian and from children themselves (where it is meaningful to do so) before taking photographs of children (under 16’s). Parent/guardians should read your information sheet and are welcome to contact the lead researcher for more information (Elaine Walklet: e.walklet@worc.ac.uk/ 01905 542420). Please note that photographs of identifiable children will not be published in the write-up of this study but will be used within the analysis.

• Please do not upload photographs that could reasonably be considered indecent. This includes photographs of nudity, sexual activity and violence.

VI Instructions and video links for accessing the online journal

Accessing your Photovoice Journal
Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: a Photovoice study

A. Accessing the journal from your computer or laptop

1. Go to https://worcesterbb.blackboard.com and type in your username and password. Enter your username and password and click “Login”. This will take you to the Blackboard home screen.
2. Under “My Organizations” click on “Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: Photovoice Journal”. This will take you to the Welcome page.
3. Scroll down the Welcome page and click on the link “Photovoice Journal”. This will take you to the journal page.
4. Click on “Create Journal Entry”. This will take you to a page to start creating your Journal.
5. Under “1. Title” type in a title (e.g. Journal entry 1).
6. Click into the large text box below the row of icons. Type in your reflective commentary about what the subject of photo 1 means to you, how it affects your quality of life and why you chose to photograph it. This part is really important for the analysis so please write as much as you feel able to – the box is expandable.
7. Click on the paperclip icon to browse to the location of the photo you wish to add.
8. Double click on the photograph which corresponds to this journal entry. Click the Submit button to attach the photo to your post.
9. Click the Post Entry button to save your post.
10. Repeat this process for your next journal entry! You can post as often as you like within the 2-week study period.
11. If you want to edit or delete an individual entry, click on the “˅” symbol next to the title of the entry you want to edit or delete. This will bring up a number of options – click on edit or delete. Note – if you click “delete” you will be asked to confirm you wish to permanently delete this entry.

B. Accessing the journal from your Apple Smart Phone/ Ipad

1. Download the Blackboard Mobile Learn app for free from your app store:

   Apple: https://itunes.apple.com/gb/app/blackboard-mobile-learn/id376413870?mt=8

2. Open up the app and tap on “Search for your school”. Type in “University of Worcester”. Tap on the “University of Worcester” link which appears. Type in your username and password and tap on “Continue” to log in.
3. Tap “Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: Photovoice Journal”.
4. Tap “Welcome” and then “Photovoice journal”. This will take you to the journal page.
5. Tap “+ Add”. This will take you to a page to start creating your journal.
6. Type in a title in the subject box (e.g. journal entry 1)
7. Tap “Attachments”. This will enable you to upload your photo.
8. Tap into the textbox and type in your reflective commentary about what the subject of photo 1 means to you, how it affects your quality of life and why you chose to photograph it. This part is really important for the analysis so please write as much as you feel able to – the box is expandable.
9. Tap “Library” to access photos you have already taken. Tap the photo you wish to upload.
   Alternatively tap “Camera” to take a new photo and upload it directly.
10. Tap “post” (next to attachments) to submit your first journal entry.
11. Repeat this process for your next journal entry! You can post as often as you like within the 2 week study period.

A video of these instructions can be accessed online: https://worc.wistia.com/medias/4qtjd2q12t

C. Accessing the blog from your Android Smart Phone/ tablet

1. Download the Blackboard Mobile Learn app for free from your app store:
2. Open up the app and click on “Search for your school”. Type in “University of Worcester”. Tap the “University of Worcester” link which appears. Type in your username and password and tap on “Continue” to log in.
3. Tap “Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: Photovoice Journal”.
4. Tap “Welcome” and then “Photovoice journal”. This will take you to the journal page.
5. Tap “+ Add”. This will take you to a page to start creating your journal.
6. Type in a title in the subject box (e.g. journal entry 1)
7. Tap “Attachments”. This will enable you to upload your photo.
8. Tap into the textbox and type in your reflective commentary about what the subject of photo 1 means to you, how it affects your quality of life and why you chose to photograph it. This part is really important for the analysis so please write as much as you feel able to – the box is expandable.
9. Tap “Library” and then “Documents” to access photos you have already taken. Tap the photo you wish to upload. Alternatively tap “Camera” to take a photo and upload directly.
10. Tap “post” (next to attachments) to submit your first journal entry.
11. Repeat this process for your next journal entry! You can post as often as you like within the 2 week study period.

A video of these instructions can be accessed online: https://worc.wistia.com/medias/uvwt18q2w9
Data Processing Agreement

Date: 12th August 2016

Parties

(1) University of the West of England, Bristol whose address is Frenchay Campus, Coldharbour Lane, Bristol, BS16 1QY, ("UWE") and
Background

1. UWE wishes the Data Processor to carry out the work detailed in Schedule 1 ("the Services").
2. Both Parties agree that they have obligations under the Data Protection Act 1998 ("the Act").
3. This Agreement records the obligations of both Parties.

Operative Provisions

Where the Data Processor processes personal data on behalf of UWE it agrees not to act in any way so as to cause UWE to breach of any of its obligations under the Act.

Specifically the Data Processor agrees to:

1. Process personal data only in accordance with instructions from UWE;
2. Process the personal data only to the extent, and in such manner, as is necessary for the provision of the Services or as is required by law or any regulatory body;
3. Implement appropriate technological measures to protect against accidental loss, destruction, damage, alteration or disclosure. Such measures shall be appropriate to the loss which might result from any unauthorised or unlawful processing, accidental loss, destruction or damage to the personal data and having regard to the nature of the personal data which is to be protected;
4. Take reasonable steps to ensure the reliability of any employees who have access to the personal data;
5. Ensure that any employees required to access the personal data are informed of the confidential nature of the personal data and comply with the obligations set out in this Agreement;
6. Ensure that none of its employees publish, disclose or divulge any of the personal data to any third party unless directed in writing to do so by UWE;
7. Notify UWE in writing (within five working days) if it receives:
   7.1. A request from a data subject to have access to that person's personal data; or
   7.2. A complaint or request relating to UWE's obligations under the Act.
8. Co-operate fully with UWE in relation to any complaint or request made, including by:
   8.1. Providing UWE with full details of the complaint or request;
   8.2. Complying with a data access request within the relevant timescales set out in the Act and in accordance with UWE's instructions;
   8.3. Providing UWE with any personal data it holds in relation to a data subject (within the timescales required by UWE); and
   8.4. Providing UWE with any information requested by UWE.
9. Permit UWE or its representatives (subject to reasonable and appropriate confidentiality undertakings), to inspect and audit its data processing activities (and/or those of its agents, subsidiaries and subcontractors) and comply with all reasonable requests or directions by UWE to enable UWE to verify and/or procure that the Data Processor is in full compliance with its obligations under this Agreement;

10. Not process personal data outside the European Economic Area without the prior written consent of IJWE and, where UWE consents to transfer, to comply with:

   10.1. The obligations of the Data Controller under the Act by providing an adequate level of protection to any personal data that is transferred; and

   10.2. Any reasonable instructions notified to it by OWE.

11. Remove all personal data from all systems of the service provider after the use of the data and provide written confirmation of this to UWE,

12. In the event that IJWE serves on the Data Processor an information notice requiring the Data Processor within such time and as required by the information notice, to provide UWE with such information as UWE may reasonably require relating to:

   12.1. Compliance by the Data Processor with the Data Processor's obligations under this Agreement in connection with the processing of Personal Data; and/or

   12.2. The rights of the data subjects, including but not limited to subject access rights.

13. The Data Processor will allow its data processing facilities, procedures and documentation to be submitted for scrutiny by UWE or its auditors in order to ascertain compliance with the relevant laws and the terms of this Agreement.

14. With respect to the parties' rights and obligations under this contract, the parties acknowledge that, except where otherwise agreed, UWE is the data controller and the Data Processor is the data processor. Where the Data Processor wishes to appoint a sub-contractor to assist it in providing the services and such assistance includes the processing of personal data on behalf of UWE, then IJWE hereby grants to the Data Processor a delegated authority to appoint on UWE's behalf such subcontractors to process personal data provided that the Data Processor shall notify UWE of the identity and location of such sub-contractors and shall seek UWE's consent in writing to such appointment. The Data Processor warrants that appointment of any sub-contractor shall be on substantially the same terms as contained in this Agreement.

15. The Data Processor shall be liable for and shall indemnify (and keep indemnified) IJWE against each and every action, proceeding, liability, cost, claim, loss, expense (including reasonable legal fees and disbursements on a solicitor client basis) and demands incurred by UWE which arise directly or in connection with the Data Processor's data processing activities under this contract, including without limitation those arising out of any third party demand, claim or action, or any breach of contract, negligence, fraud, wilful misconduct, breach of statutory duty or non-compliance with any part of the Act by the service provider or its employees, agents or sub-contractors.

16. No term of this Agreement shall be enforceable under the Contracts (Rights of Third Parties) Act 1999 by a third party, but this does not affect any right or remedy of a third party which exists or is available apart from under that Act.

17. This Agreement shall be governed by and construed in accordance with English law.
Each of the Parties irrevocably submits for all purposes in connection with this Agreement to the exclusive jurisdiction of the courts of England.

Authorised Signatory University of the West of England, Bristol (Data Controller)

Name: Larry Rawlinson
Title: Head of Contracts
Date:

Authorised Signatory:

Name: Elizabeth Symonds
Title: Team Leader, Learning and Teaching Technology Unit (Data Processor)
Date: 16/8/16

Schedule 1 — The Services

Elizabeth Symonds will receive participant names and email addresses via secure internal University of Worcester email from the lead researcher at UWE, Bristol ie Elaine Walklet.

All names and email address will be stored securely by Elizabeth who will use them to set up participants with a log-in for a University of Worcester hosted Blackboard page where they will have access to a private online journal. Participant's accounts and individual journals will be set up to be anonymous.

Elizabeth Symonds and team (Dave Wager, Pete Thornton and Gerry Beattie) will have access to anonymised journals due to being the site administrators. In the event a participant wishes to withdraw from the study, Elizabeth Symonds will permanently delete their account.

VIII Patient Partner Agreement and Signed Letter of Understanding

Confidentiality Agreement relating to
Public Research Partner participation in University Research*

Between

Karen Butcher and

University of the West of England, Bristol

*This agreement is a legally binding document. You are advised, but not obliged, to seek independent legal advice where you feel it appropriate to do so.

CE-16-0268

Confidentiality Agreement between

1. Karen Butcher of XXXX; and
2. University of the West of England, Bristol of Frenchay Campus, Coldharbour Lane, Bristol, BS16 IQY ("UWE").

It is hereby agreed as follows:

I. In consideration of my participation as a Public Research Partner in the Study entitled 'Quality of life in adults with Charcot-Marie-Tooth (CMT) disease: a Photovoice study" I understand that I may receive confidential information and/or Personal Data (as that term is defined under the Data Protection Act, 1998) which is required to be maintained in strict confidence.

II. I hereby undertake that in respect of any confidential information and/or Personal Data which I may receive, I shall:

(a) receive and keep any confidential information including any Personal Data secret and confidential and not disclose such confidential information to any third party;
(b) use the confidential information only for purposes of carrying out my role as Public Research Partner;
(c) only disclose or discuss the confidential information to those individuals (including other Public Research Partners) as formally approved by the UWE Project Manager or other identified Project Lead and who have a need to have access thereto wholly necessarily and exclusively for the purposes of the Study which may include but not by way of limitation any legal/statutory/regulatory or other proceedings in relation to which I may be required to make a disclosure;
(d) not make any personal or commercial use of or make any personal or commercial gain from the confidential information or seek to obtain any protection of the intellectual property contained in the confidential information.

III. Upon notification of termination of the Study and/or my involvement in the same, I will immediately return any confidential information and any copies thereof in my possession or under my control and make no further use or disclosure of any of the confidential information. If UWE so dictates, the confidential information shall be destroyed.

IV. I acknowledge and agree that the intellectual property and copyright in confidential information disclosed to me, including any documents, files and any other items containing any confidential information belongs/or is under licence to UWE. It will not be given to any other persons or parties nor removed from UWE premises unless expressly requested or consented to by UWE.

V. This Agreement shall neither prejudice nor limit the rights of UWE in respect of any intellectual property rights in the confidential information.

VI. I shall not arrange nor permit the publication of any information regarding the results or outcome of the confidential information or the Study without the prior written consent of UWE.

VII. I understand and accept that UWE gives no warranties in relation to any information, confidential or otherwise, disclosed to me in pursuance of my role as Public Research Partner and in particular (but without limiting the foregoing) no warranty or representation, express or implied, is given by UWE as to the accuracy, efficacy, completeness,
capabilities or safety of any materials or information as I may receive as a Public Research Partner in the Study.

VIII. By signing this Confidentiality Agreement I agree to be legally bound by the terms herein. I understand that I am free but not obligated to seek independent legal advice regarding my obligations hereunder.

IX. Except in certain circumstances, where UWE deems that confidential information disclosed by me to UWE is required to be disclosed to a third party, which may include but not be limited to instances where my safety or the safety of another is deemed to be at risk, UWE hereby undertakes to maintain in confidence any information disclosed by me to its staff where the confidentiality of the information is made known by me at the time of disclosure or where the information is Personal Data as defined under the Data protection Act.

X. This Confidentiality Agreement will be either come into effect upon its signature or will be deemed to have come into effect upon the first date upon which I received any confidential information or Personal Data in respect of the Study where that date predates signature, and by signing this document, I warrant and confirm that I have not to date caused any breach of this undertaking by unlawfully disclosing any confidential information or Personal Data already received by me.

XI. By signing this Agreement, I confirm that I have read, understood and agree to be bound by UWE's Policy on Data Protection as though I were a member of staff which is available at http://wwwl.uwe.ac.uk/aboutus/policies

XII. The terms of this Confidentiality Agreement shall endure in perpetuity unless and until any confidential information is published or otherwise disseminated with the consent of the University of the West of England, Bristol.

Signed By Karen Butcher

____________________________
signed Karen Butcher

____________________________
print name Chief Operating Officer

____________________________
title 08/08/16

____________________________
date

Signed on behalf of the University of the West of England, Bristol
signed Larry Rawlinson print name Head of Contracts title

18/10/2016 date
Contributing to research at UWE as a Public Research Partner

Letter of understanding

Public involvement in research is where research is being carried out 'with' or 'by' patients, service users or members of the public rather than 'to', 'about' or 'for' them. Being involved might mean contributing to research in a variety of ways, drawing on personal experiences. For example, of having a health or other problem, of using services or having treatments, or bringing a particular perspective — as an older person or as a member of a particular social or ethnic group. The work might include helping to identify research priorities, contributing to the planning and conduct of research, and/or supporting the sharing of research findings and getting evidence into practice.

When you contribute to research at UWE as a Public research Partner you can expect:

1. To be treated with respect, dignity and equality.
2. To have an appropriate identified contact within a research study who will answer questions and provide support.
3. To receive clear and accessible information about the research.
4. To be advised on maintaining confidentiality of University or of other party information and of your obligations under the data protection legislation to maintain confidentiality of any personal data you may learn of, some of which may be sensitive. You will be asked to sign an agreement that confirms your understanding of this and your agreement and commitment to maintaining all such matters confidentially.
5. To have a good understanding of how you might contribute, including the different tasks you might help with, acknowledging that the role might change and develop over time.
6. To negotiate ways of working with researchers, including discussions on any reasonable adjustments to ways of working we may be able to make on your behalf, in order to fully support you in your role. This may include, amongst other things, giving you time out from your involvement as needed, and any other support you may require should your involvement cause you any discomfort or distress.
7. Volunteer: To have any agreed out of pocket expenses covered.
8. To have feedback on your contributions from the researchers on the project with which you are involved, and to have your contributions actively considered in making changes to, and decisions about, research.
9. To raise any concerns or issues with researchers initially, and to have the contact details of a UWE staff member outside the research project who you can talk to if there are any problems, or should you wish to make a complaint. For your research project the contacts are Elaine Walklet (e.walklet@worc.ac.uk) within the research team and Andy Gibson (Associate Professor in Patient and Public Involvement) (andy.gibson@uwe.ac.uk) outside the research team.
10. Some involvement roles might need other forms to be completed. For example, if you will be doing interviews with research participants you may need an honorary contract and/or a Research Passport which are standard documents allowing access to facilities, patients, volunteers and staff of UWE and/or other organisations such as the NHS. This will ensure that your activity is covered by the appropriate indemnity insurance scheme. For some roles you may need a Disclosure and Barring Service (DBS) check and to have completed safeguarding training. If these formalities are required you can expect researchers to explain the processes needed and to be supported with form filling and any agreed costs.

11. To have a proportionate induction which addresses the above issues and provides you with the information, expenses forms etc. that you need, including information about relevant UWE Policies and guidance.

When you are involved in research at UWE researchers will expect the following from you:

1. That you will have relevant personal experiences for the specific research you are to be involved in.

2. To treat researchers, colleagues and peers and their information and data with dignity and respect and in particular to ensure that UWE’s policies on the same are upheld and followed at all times. UWE’s equality and diversity policy can be found here:

http://wwwl.uwe.ac.uk/aboutus/visionandmission/equalityanddiversity/policiesandprocedure s.asp x

3. To share relevant personal experience and contribute in the ways agreed.

4. To let researchers know if the involvement role is not working for you, and let UWE know if you feel you have not been treated appropriately.

5. To let researchers know if you do not wish to continue to be involved, or if you need to take time out.

6. To maintain confidentiality and data protection in line with UWE policy, contracts with funders and any other specific agreements which will require you to sign a further agreement and to warrant that you will not take any information, research data or other data including but not limited to personal or other sensitive data away from UWE premises in any format, including not downloading research data to a non-UWE computer or device, unless specifically required to do so by UWE and that you can evidence how security of the same may be maintained.

7. To attend any relevant training as might be necessary for you to be able to carry out your role in accordance with all laws, regulations and/or UWE policies and procedures. Such UWE procedures include but not by way of limitation the UWE Code of Good Research Conduct, Research Data Guidelines, the Policy on Good Research Conduct, available here:

http://wwwl.uwe.ac.uk/research/researchgovernance/codeofgoodresearchconduct.aspx

UWE very much appreciates the important contributions that members of the public make to research. If you would like to be involved in research at UWE and understand what UWE will offer and what is expected from you, please will you sign one copy of the letter and return it to Elaine Walklet.
With best wishes

Elaine Walklet

I understand what being involved in research at UWE means and I understand the agreement described in this letter:

Karen Butcher

(print name)

Signed on behalf of UWE by: Larry Rawlinson æ-A-20 <

Signed by UWE Research Project Manager:

ELAINE WALKLET

(print name)

signature
IX Data management plan

DMP title

Project Name Quality of life in adults with CMT: a photovoice study

Principal Investigator / Researcher Elaine Walklet

Project Data Contact Elaine2.Walklet@live.uwe.ac.uk

Institution University of West England

Data Collection

What data will you collect or create?
Personal details - Participants’ names, email address, basic demographic details (e.g. age, sex), type of CMT/ any additional health issues

Participant generated photographs and diary entries submitted to a specifically designed, secure online Blackboard site (Storage capacity = unlimited, Back-up provision = Blackboard is fully backed up and held on a secure server by Blackboard Managed Hosting).

How will the data be collected or created?
Participants expressing an interest in the study will be sent a copy of the participant information sheet and consent form via email. In addition, a web link and password will be provided to access a Qualtrics survey which will contain the information sheet, online consent form and online personal details form.

Photographs and diary entries will be collected via a specifically designed, secure online Blackboard site. Each participant will have access to an individual private journal which will be accessible to them, the lead researcher and the UoW learning technologists administering the site. Each journal will be set up with an anonymised log-in and a participant generated pseudonym.

Documentation and Metadata

What documentation and metadata will accompany the data?
Participant details (e.g. name, email address, demographics) will be stored in a single excel file labelled "Quality of life in adults with CMT_Participant details". This will include the date that participation commenced and the 2 week completion date.

Anonymised diary entries and photographs will be downloaded and stored in journal order (e.g. journal entry 1: photograph and text, journal entry 2: photograph and text). Each participant will have a separate file saved under their pseudonym. These will be called "Quality of life in adults with CMT Xxx".

Ethics and Legal Compliance

How will you manage any ethical issues?
Full ethical approval will be obtained from UWE. Informed consent will be obtained for data preservation and sharing (in addition to all other aspects of the study). Data will be anonymised to protect the identity of participants.

How will you manage copyright and Intellectual Property Rights (IPR) issues?
As a student on the Prof Doc in Health Psychology course, Elaine Walklet will own the IPR. Participants will be required to consent to the publication of any submitted photographs (except photographs of identifiable people).
Storage and Backup

How will the data be stored and backed up during the research?
Personal information will be stored online within the European Economic area (password protected account, servers automatically backed up) and within a password protected microsoft excel file, saved on a secure University of Worcester network. This is due to the researcher being employed on a full-time basis at the University of Worcester. The excel file will be manually backed up on an encrypted memory device.

Photographs and diary entries will be stored on a password protected Blackboard site (Blackboard is fully backed up and held on a secure server by Blackboard Managed Hosting). For the purposes of analysis, anonymous written diary entries and photographs will be copied into password protected microsoft word file immediately after the 2 week study period. These will be stored on a secure University of Worcester network and backed up manually on an encrypted memory device.

How will you manage access and security?
Passwords to the secure online platforms will be shared with the UWE research team in the event that primary data needs to be accessed.

Selection and Preservation

Which data are of long-term value and should be retained, shared, and/or preserved?
All data will be retained for up to 5 years.

What is the long-term preservation plan for the dataset?
Following dissemination, data will be stored on a secure University of Worcester drive

Data Sharing

How will you share the data?
Identifiable information will be accessed by the lead researcher only. Contact details (name, email address) for setting up the journal will be shared with the lead learning technologist with permission from participants. Non-identifiable information (e.g. anonymised photographs and diary entries) may be shared within the research team during the analysis. In this instance data will be shared securely using the Blackboard site for the study. Anonymised quotations and photographs will be disseminated via conference presentations, peer-reviewed articles and within a doctorate thesis.

Are any restrictions on data sharing required?
Consent will be obtained to use photographs and verbatim quotes in dissemination activities.

Responsibilities and Resources
**Who will be responsible for data management?**
Elaine Walklet will be responsible for data management. All members of the research team including the patient partner agree to abide by the UWE Code of Good Research Conduct.

**What resources will you require to deliver your plan?**
All members of the research team (including patient partner) agree to abide by the UWE Code of Good Research Conduct. No additional resources or training are required.
Appendix C: Analysis

I Identifying my own perspective

HRQoL to me = Being able to do things I value e.g. relationships, achieve and contribute to society (work), experience new things (travel), maintain my functioning.

+ James – practical help and reassurance + family who understand/ support and friends
+ Car and disabled badge – means I can get to places
+ Being stubborn (but this can also be negative)
+ Exercises – pilates and physio
+ Adoptions and devices to help functioning (but can be restrictive)
+ Medication
+ Surgery (but complications and interruption to life are negative)

- People not knowing I have a condition or understanding what it is because nobody has heard of it. Not wanting to label myself as disabled and wanting to be seen as normal but also needing people to understand when I can’t do things.

- Pain – every day and reliant on painkillers

- Every day things take me longer or require assistance. Particularly because of my hands but also lack of functioning in my legs. Causes anxiety – what if I fall? Will I be able to sit down? How bad will it get?

- Work-life-condition management balance – appointments + management takes up lots of time (plus everything takes me longer) so I can’t fit everything in. Constantly battling with myself and feeling guilty I am neglecting something.
21st May 2017

Two thought provoking events occurred this week. Firstly, two participants who (presumably) do not know each other posted about the same thing (the usefulness of having a ‘blue badge’) within 15 minutes of each other. This surprised me as I think it must be quite unlikely to decide to talk about the same issue on the same day and the same time! It was also really reassuring as it highlighted that the things that participants are writing about are clearly shared experiences. The other thing that happened which was quite profound was that one participant posted their first entry with quite a brief comment. I started trying to interpret what the picture was about from the brief description and thought I could make some sense of it. Later, I found the participant had followed the guidance for what to write about and had written a lot more as a comment, which gave the image a completely new meaning. This highlighted to me that the written interpretations of participants are really important. It also made me think about the way that participants interpret meanings and whether part of this happens during the writing process itself.

22-23rd May 2017

I attended a 2-day NVivo workshop and found it so helpful. Initially I was unsure why the training was 2 days and worried it might be a waste of time. I think on reflection however it did need to be two days as there was so much to learn and the training focused on helping you to work out how you could make best use of it. I found the individualised approach to the training particularly useful as it helped me to work out how I am going to analyse my data. This was a huge relief and I can’t wait to get started!
30th May 2017

Today I met with Tim to talk through our independent coding of one set of journal entries. I found this a really helpful experience, not least because it was useful to talk through the data together. I realised that we were both picking up on similar concepts but the language I was using to label the codes was a little descriptive, and not always reflective of the meaning I attached to the code itself. I will go back and review my coding in light of this. We also discussed how the journal left both of us feeling a bit sad and the importance of stepping away from the data after working on the analysis. Some of the experiences illustrated are quite close to my own and we talked through how this affected my interpretation, in particular my interpretation of the images. For example an image of an uneven pavement had little emotional associations for Tim but made me feel quite anxious! Talking through my interpretations was really helpful and I will continue to do this throughout the analytic process.

17th July 2017

I think it’s starting to come together now which is a huge relief as I have felt a bit lost in it all recently. Everything seems so important and inter-connected! I have developed clear themes but I’m not convinced about the titles and there are a few aspects I am unsure of. I’m hoping it will help to talk through my initial analysis with Eleanor later this week and then think through how best to present it. There is so much data to choose from; I feel really privileged to be analysing it and I just hope I am able to do it justice.