Coping strategies used by children living with a genetic muscular disorder in New Zealand

Aaron Anand

A thesis submitted to Auckland University of Technology in partial fulfilment of the requirements for the degree of Master of Health Science (Psychology)

Department of Psychology
2019

Abstract

Objective: Children living with a genetic muscle disorder (GMD) report significant impairments in health-related quality of life (HRQOL) as a result of living with a chronic and progressive disorder. The challenges posed by living with a GMD may lead to additional and increasing daily stressors for these children and their families/whanau. Children use a range of coping strategies in an effort to respond to such stressors. Currently, however, limited literature is available regarding the use of coping strategies in children living with a GMD. The literature that is available focuses predominantly on a single GMD - Duchenne muscular dystrophy. This study sought to explore self-reported use and perceived effectiveness of coping strategies among children across a range of GMD and to identify associations with HRQOL.

Method: The study examines secondary data collected as part of a nationwide, epidemiological study of the prevalence and impact of GMDs in NZ (MD-PREV). This sub-study involved 48 children (aged 5 to 15 years); 29 (60.4%) were male participants. Children were invited to complete the Kidcope and Paediatric Quality of Life questionnaires as part of the initial impact assessment. Inclusion in this sub-study required a clinical confirmation of a GMD, this included muscular dystrophy, congenital myopathy and ion channel muscle disorders.

Results: Cognitive restructuring (n = 34) and wishful thinking (n = 34) were the most commonly used coping strategies, whereas, social support (*a lot,* 75.00%), resignation (*a lot,* 55.60%), and distraction (*a lot*, 50.00%) were perceived as the most effective coping strategies. Positive coping strategies (M = 2.36, SD = .64; $p < .01$) were perceived to be more effective compared to negative coping strategies (M = 2.07, SD = .77; $p < .01$). There were no significant associations between the types of coping strategies used and children's age, sex, or ethnicity ($p > .05$). Total number of positive ($p < .01$) and negative ($p < .01$) coping strategies used, along with ethnicity ($p < .05$), significantly accounted for 49% of total variance observed in self-reported HRQOL ($p < .01$). Sex and age were also included in the model, however, did not contribute significantly to variance seen in HRQOL ($p > .05$). Lower total HRQOL was significantly associated with the increased use of both positive and negative coping strategies ($p < .01$)

Conclusions: Study findings suggest that children use a range of positive and negative coping strategies when living with a GMD. While children reported positive coping strategies as more effective than negative strategies, results suggest that childrens use of a greater number of different coping strategies is associated with poorer self-reported HRQOL. Children living with a GMD may benefit from psychoeducation around how to use coping strategies effectively. Increased education for children, and their families/whanau, may help them to better cope with stressors related to their health condition.

Table of Contents

List of Figures

List of Tables

Attestation of Authorship

I hereby declare that this submission is my own work and that, to the best of my knowledge and belief, it contains no material previously published or written by another person (except where explicitly defined in the acknowledgements), nor material which to a substantial extent has been submitted for the award of any other degree or diploma of a university of institute of higher learning.

Signed: 

Acknowledgements

I would like to give a big shoutout to everyone who has helped me enormously throughout this long journey. Firstly, I would thank my supervisors Kelly and Charmaine. Both of you have been instrumental in helping me through this challenging year, could not have been possible without all your help. Thank you.

Secondly, I would also like to thank all my friends and family for their never-ending support throughout the years. Sheryl, Sharonrose and Mum for never letting me give up, Charlie for providing the sold banter (and snacks), Jermaine for providing the beats, Jake for the emotional support, and last but not least Mr. Morris for being my number one supporter.

I would like to thank my partner for being the best all-rounder, thank you for being my proof-reader, motivator, trainer, procrastination buddy, personal chef and so much more.

Ethics Approval

The current study was approved by the Auckland University of Technology Ethics Committee (Reference number: 14/296) on the 14th of September 2014 (see Appendix A).

Chapter One: Introduction

Genetic muscular disorders (GMDs) are a diverse group of hereditary genetic disorders that commonly present with progressive muscle weakness, causing significant comorbidities and as a result reduce an individual's daily functioning (Theadom et al., 2019). GMDs impact several domains of an individual's health including physical, psychological and social domains (Jones et al., 2018). It is widely shown in the literature, that a substantial proportion of people living with a chronic illness are affected by a type of GMD (Theadom et al., 2019; Norwood et al., 2009). Zatz, Passos-Bueno and Vainzof (2016) indicated GMDs affect over 1 in 1,000 individuals worldwide. A New Zealand (NZ) based study identified 966 cases of confirmed clinically diagnosed individuals with GMDs living in NZ (4,242,048), attained though the 2013 NZ population census (NZ Statistics, 2013) (Theadom et al., 2019). Several studies have identified significantly increased pressure on close family members, both physically and psychologically (Jones et al., 2018).

This thesis will explore the use of coping strategies in children with a wide range of GMDs and the impact this has on their health-related quality of life (HRQOL). This first chapter will provide a brief overview on the statement of the problem, the aims of the current study, and its significance.

1.1 Statement of the problem

As mentioned above, GMDs are a diverse group of hereditary genetic disorders that commonly present with progressive muscle weakness, often causing significant health impairments and negatively impacting an individual's daily functioning. Subsequently, negatively impacting their HRQOL (Theadom et al., 2019). GMDs encompass a range of neuromuscular disorders including but not limited to muscular dystrophies, myopathies, motor neuron diseases, ion channel diseases, and neuromuscular junction diseases (Huml, 2015; Jones et al., 2018; Lovering et al., 2005; Theadom et al., 2019). These disorders are often the result of mutations in one or more genes that control normal muscle structure and functioning in an individual's genetic code (Abbott & Carpenter, 2015). For example, Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are caused by

mutations in a gene in the X chromosome. GMDs are classified by clinical presentation, mode of inheritance, age of onset, and overall progression of symptoms (Huml, 2015; Lovering et al., 2005). Disorder progression varies, for example boys with DMD commonly need a wheelchair by the age of 12, as their limbs and spine become progressively deformed, whereas children with BMD may struggle with climbing the stairs at the age 12 (Webb et al., 2005). Along with experiencing several comorbidities, including, but not limited to cognitive, cardiac, respiratory, orthopaedic complications and cognitive disabilities (Jones et al., 2018; Lovering, Porter, & Bloch, 2005).

Life expectancy for GMDs is significantly lower compared to individuals without a GMD. This significantly differs on the type and severity of the GMD. The life expectancy for DMD is early 30s and BMD is mid-40s. Unfortunately, there are no cures for GMDs. However, due to continued advances in supportive medical care children are now living longer. Treatment is most commonly focused on symptom management to optimise quality of life (QOL) and functioning (Jones et al., 2018; Kohler et al., 2009; Strehle et al., 2009).

Zatz and colleagues (2016) suggested neuromuscular disorders (which includes a wide group of genetic conditions) affect 1 in 1,000 individuals worldwide. In Northern England, Norwood and colleagues (2009) found the combined population prevalence figure for all inherited muscle disease categories in their clinic database was 37.00 per 100,000 persons. Therefore, concluding that although several of the identified conditions represented individually may be rare, however, cumulatively they are an important proportion of those with chronic disease. In NZ, point prevalence of all genetic muscle disorders was 22.3 per 100,000 (95% CI 19.50–25.60); and 20.92 children (5-14 years) per 100,000 (95% CI 17.42–25.11) in NZ (Theadom et al., 2019).

Children with a GMD can start to experience muscle weakness as early as three years old. Initial effects are common in the muscles of the hips, pelvic area, thighs and shoulders. This can later progress to affect the skeletal muscles in the arms, legs and trunk (Genetics Home Reference, 2019; Huml, 2015; Muscular Dystrophy Association, 2019). Skyrme (2017) and Morris (1992) suggested that children with GMDs, are physically limited by pain, discomfort, increased muscle weakness, and a lack of energy. However, children are not only impacted physically, but there are also negative psychosocial and cognitive effects of GMDs

(Jones et al., 2018; Lovering et al., 2005). For example, Skyrme (2017) found that alongside such physical symptoms, affected children report increased feelings of isolation, discrimination and a range of HRQOL issues. Similarly, Wei, Speechley, and Campbell (2015) identified a significant association between wheelchair use and poorer HRQOL.

There is a gap in the literature when considering how children cope with symptoms commonly associated with GMDs and how this may impact children's HRQOL. It is essential to assess coping in children with serious and chronic illness to provide a better understanding of the impact the illness has on the child. To date, there is little literature looking at the impact of coping strategies and children's self-reported HRQOL in GMDs populations. An extensive literature review on the underpinning framework of coping strategies and use in paediatric population will be discussed in chapter 2, however there is very limited literature when considering the use and effectiveness of coping strategies in GMD children populations. Previous child studies utilising the Kidcope found most successful coping strategies were active and passive coping strategies. For example, Smith and colleagues (2013) concluding adolescents most often use cognitive-oriented coping strategies in spinal cord injury (SCI).

There is a lack of evidence when considering HRQOL across the broader spectrum of GMDs, despite the variation in the muscles affected, severity, age of onset, and nature of progression (Landfeldt et al., 2015; Lue et al., 2016). Further evidence is needed on the coping strategies children utilise in managing the implications of living with a genetic muscle disorder.

1.2 Current study aims

This research aims to identify the types of coping strategies commonly used by children with GMDs, the efficacy of these strategies, and how they may differ depending on the individual characteristics of the child. In addition, the research aims to examine the relationship between children's use of coping strategies and thier self-reported HRQOL.

The key study aims were:

1. To identify the types of coping strategies used by children living with GMDs and the effectiveness of these strategies.
2. To understand what extent of children's use of coping strategies differ by their age, sex, and/or ethnicity.
3. To determine what associations exist between the types of coping strategies used by children and their self-reported HRQOL.

1.3 Significance of the current study

Study findings may inform future research and the development of new interventions to support children living with a GMD and their families. This study will provide crucial information on commonly used coping strategies in children with GMDs, which in turn, will provide further information on the types of coping strategies that children living with a GMD find to be helpful. Therefore, this study will highlight key areas for future research and identify adaptive coping strategies that may positively impact HRQOL of children with GMDs. This study also adds to the present body of literature comparing coping strategies utilised by children. For example, in terms of clinical impact, study findings suggest that particular types of coping strategies are useful for this population. Clinicians working with children with GMDs could then recommend that children try using alternative or additional coping strategies in an effort to improve wellbeing. Research in this area is especially important given there are currently no cures for GMDs. Identifying the most effective coping strategies and those linked to better HRQOL outcomes may help to inform the types of support and recommendations offered to children facing the daily challenges of living with a GMD and their families/whanau.

1.4 Outline of thesis

This thesis will consist of 5 chapters. Chapter One provides a brief introduction, which consists of a statement of the problem with context to the impact GMDs have on children, the aim of the current project, and the significance of presenting the findings from this study.

Chapter Two is a comprehensive review of the current literature. Firstly, it defines GMDs, followed by a brief insight into the pathophysiology, prevalence, impact on HRQOL and available treatment. The second part of Chapter Two is an in-depth review of relevant literature regarding the development of different types and use of coping strategies by children. Lastly, Chapter Two will briefly discuss the use of a bio-behavioural model in children with a GMD.

Chapter Three describes the quantitative methodology and methods of the current study, including a brief insight into the larger nationwide, epidemiological study of the prevalence and impact of GMDs in NZ (MD-PREV) study. The study design, participant demographic information and statistical analysis will also be outlined in Chapter Three.

Chapter Four will presents the findings from the current study in relation to each stated aim. This will be presented in the form of tables and figures, with a brief overview of the key findings.

In the final chapter, the findings of the present study have been discussed in relation to previous literature. The implication of the current study's findings have also been discussed. The strengths and limitations of this study, along with recommendations and directions for future studies will also be discussed in Chapter Five.

Chapter Two: Literature Review

This chapter will provide a general overview of GMDs, including its definition, prevalence, pathophysiology, risk factors, treatment, and impact on affected children and their families. It will provide a critical review of the current literature regarding the types of coping strategies that are commonly used by children in a range of health conditions (such as, GMDs and SCI) and psychological situations (such as adaption). It will explore the impact of GMDs on HRQOL and the influence of coping strategies on HRQOL. The potential use of a bio-behavioural model in children populations will be discussed

2.1 Genetic muscular disorders

This section will provide an overview on GMDs, with specific examples of how they differ in their presentation and progression across the different disorders. This will be followed by a review of GMD prevalence rates, both globally and in NZ. A brief overview of the pathophysiology, risk factors and available treatment will be discussed in the current section.

2.1.1 Definition

As mentioned in the previous section, GMDs commonly present with progressive muscle weakness, causing significant comorbidities and as a result reduce an individual's daily functioning (Gazzard, 2004; Theadom et al., 2019). They are often classified by clinical presentation, mode of inheritance, age of onset, and overall progression of symptoms (Huml, 2015; Lovering et al., 2005). GMDs include a range of neuromuscular disorders including muscular dystrophies, myopathies, ion channel diseases, neuromuscular junction diseases mitochondrial diseases, and motor neuron diseases (Huml, 2015; Jones et al., 2018; Lovering et al., 2005; Theadom et al., 2019).

Muscular dystrophies are a group of disorders that cause degeneration and weakness of the skeletal muscles, common examples include DMD and BMD (Muscular Dystrophy Association, 2019). Congenital myopathies are a group of GMDs clinically characterised by hypotonia and weakness, usually present from birth, and often have a static or slowly progressive clinical course (Cassandrini et al., 2017; Todd et al., 2018). Myopathies cause

muscle fibres to function improperly as a result of the tone and contraction of skeletal muscle, resulting in muscular weakness. Common examples include central core and multiminicore myopathies associated with ryanodine receptor 1-related (RYR1) mutations (Cassandrini et al., 2017). Ion channel disorders are associated with defects in proteins that control ion channels, this is often marked by muscular weakness, absent muscle tone, and/or episodic muscle paralysis (Genetics Home Reference, 2019; Muscular Dystrophy Association, 2019). These difficulties place children at higher risk for medical complications, including cardiac, respiratory and orthopaedic complications (Jones et al., 2018; Lovering et al., 2005) and cognitive disabilities (Jones et al., 2018).

2.1.2 Prevalence

Zatz and colleagues (2016) suggested neuromuscular disorders (which includes a wide group of genetic conditions) affect 1 in 1,000 individuals worldwide. In Northern England, Norwood and colleagues (2009) found the combined population prevalence figure for all inherited muscle disorders in their clinic database was 37.00 per 100,000 persons. Norwood et al. (2009), therefore, concluded several of the identified GMDs represented individually may be rare in the population, but accumulative with all GMDs, they combine to produce a significant proportion of those with chronic disorders in the general population. In NZ, the age-standardised minimal point prevalence of all GMDs across all ages is estimated at 22.3 per 100,000 persons (95% CI 19.5–25.6); and 20.92 for children (5-14 years) per 100,000 (95% CI 17.42–25.11) persons. (Theadom et al., 2019). Norwood and colleagues (2009) suggested previous literature are likely to underestimate those diseases that lead to early death, such as DMD, as prevalence reflects both incidence and duration (Norwood et al., 2009).

Previous studies examining the prevalence of GMDs offer a range of estimates from 28.60 per 100,000 (1 in 3,500) (Emery, 1991), 35.50 per 100,00 (Hughes, Hicks, Nevin & Patterson, 1996), and 53.10 per 100,000 persons (Darin and Tulinius, 2000). This is in contrast to Norwood and colleagues (2009), who concluded previous literature are likely to underestimate. Of note, these studies included patients with relatively common hereditary motor and sensory neuropathies, metabolic myopathies, mitochondrial myopathy and X-linked bulbospinal neuronopathy, motor neurone diseases and familial myasthenia gravis.

Each of these conditions was excluded from prevalence estimates offered by Theadom et al. (2019) and Norwood et al. (2009). Each of these conditions was also excluded from the current study, as outlined in chapter three.

Importantly, there appear to be ethnic disparities in the prevalence of GMDs in NZ. The prevalence in GMDs among NZ Europeans is estimated at 24.4 per 100,000 persons (95% CI 21.1–28.3). This rate is double that observed in the other three main ethnic groups in NZ; Māori (12.6 per 100,000 persons), Pasifika (11.0 per 100,000 persons), and Asian (9.13 per 100,000 persons). Theadom et al. (2019) concluded that in NZ individuals of European descent experienced a higher prevalence of GMDs compared to all the other key ethnic groups.

2.1.3 Pathophysiology

Given the present study encompasses a broad range of GMDs, this section will provide a broad overview of the pathophysiology of GMDs. A GMD is a condition that can be present at birth (congenital, often resulting from a mutation or mutations in the encoding genes), yet symptom onset may be delayed until later in an individual's lifetime (Webb et al., 2005). For example, DMD onset of symptoms is commonly observed in children aged between 2 to 5 years (Lovering et al., 2005; Webb et al., 2005). However, the mutation is present from birth and is an X-linked recessive disorder, most commonly present in males. In central core and multiminicore myopathies, the mutation is present at birth and is associated with RYR1 (Cassandrini et al., 2017). Generally, the mutation is in a gene that impacts the product of a specific protein (Genetics Home Reference, 2019; Lovering, et al., 2005; Zatz et al., 2016). GMDs may be X-linked recessive, autosomal recessive, or autosomal dominant (Gazzard, 2004, Lovering et al., 2005).

Importantly, the pathophysiology of GMDs can be similar and/or vary across conditions. For example, DMD and BMD are both caused by the lack of the protein dystrophin (Webb et al., 2005). Dystrophin can be described as a rod-shaped cytoplasmic protein, which is an essential part of the protein complex, it connects the cytoskeleton of a muscle fibre to the surrounding extracellular matrix through the cell membrane. Emery (2002) described this complex as the dystrophin-associated protein complex or costamere.

Dystrophins key role is to support muscle fibre strength. A lack of the dystrophin protein in muscle cells can causes them to become fragile and as a result, easily damaged (Emery, 1987; Muscular Dystrophy Association, 2019). Therefore, the absence or impairment of the dystrophin affecting the formation and maintenance of healthy muscles which leads to progressive muscle wasting, which eventually results in severe impairment (Emery, 1987; Muscular Dystrophy Association, 2019). It may either cause complete impairment in the production of the dystrophin protein (such as in DMD) or the genetic mutation may cause partial impairment in dystrophin (as seen in BMD) (Lovering et al., 2005; Huml, 2015). Therefore, DMD tends to be associated with faster and more severe progression compared to BMD (Genetics Home Reference, 2019; Huml, 2015; Lovering et al., 2005).

In contrast, congenital myopathies related to the RYR1 include a group of genetically and phenotypically diverse neuromuscular disorders (Todd et al., 2018). In the United States, it is approximated RYR1 related congenital myopathies affect 1 in 90,000 paediatric individuals (Amburgey et al., 2011). In brief, the pathophysiology of congenital myopathies significantly differs from muscular dystrophies. The RYR1 gene codes transmembrane calcium ion channels, which are embedded within the sarcoplasmic reticulum of skeletal muscle and plays a crucial role in the contraction coupling mechanism, known as excitation and contraction (Todd et al., 2018; Zalk et al., 2014). Incorrect encoding of RYR1 variant causes either chronic calcium ions leaking or over release of calcium ions from the sarcoplasmic reticulum (Todd et al., 2018; Zalk et al., 2014).

National Institute of Neurological Disorders and Stroke (NINDS) (2019) defined congenital myopathies as a group of GMDs that are often characterised clinically by hypotonia and weakness, usually present from either birth or infancy (Cassandrini et al., 2017). Although muscular dystrophies are also genetic disorders causing muscle weakness, they differ from congenital myopathies in the age of onset, distribution of affected muscles, and the presence of characteristic dystrophic changes (such as muscle fibre necrosis and regeneration) not present in biopsy specimens from congenital myopathies (Rubin, 2019; NINDS, 2019). There are several sub diagnoses of congenital myopathies, including nemaline myopathy and central core disease. Congenital myopathies are associated with several complications ranging from mild to severe, such as delayed motor milestone achievements, scoliosis, pneumonia, respiratory failure, feeding problems and ultimately death (Todd et al., 2018). Congenital myopathy diagnosis is characterised by clinical

findings, the diagnosis is often confirmed by conducting a muscle biopsy and sometimes a magnetic resonance imaging (MRI) is required (NINDS, 2019). In contrast, central core disease diagnosis is often characterised through oxidative histological staining, this process will reveal the centralised absence of mitochondria, this is seen as non-stained areas known as '*cores*' on a muscle biopsy. Cores can span the length of an entire muscle fibre (Todd et al., 2018). The most common congenital myopathy is nemaline myopathy, this is characterised by toddlers have difficulties with breathing and feeding early on. Which may later result in skeletal problems, such as scoliosis (NINDS, 2019). In general, the weakness does not worsen during the lifetime.

2.1.4 Impact of GMDs on children's Health-related Quality of Life

HRQOL in one of the most commonly used health-related concepts as it provides a multidimensional approach to examining an individual's wellbeing (Dobkin, 2004; Kawecka-Jaszcz, 2013; Law et al., 2014). HRQOL is subjective and often difficult to define; however, it is generally considered a multidimensional construct, consisting of three broad domains: physical, mental and social (Opara & Jaracz, 2010; Hunt et al., 2016; Law et al., 2014). Often HRQOL is examined by researchers to gain insight into an individuals' perception of their wellbeing in the context of an illness and/or treatment (Opara & Jaracz, 2010). Previous literature has often argued the importance of viewing health as a multidimensional component, which encompass all dimensions of health including human functioning and wellbeing, instead of just the biological dimension (Hunt et al., 2016; Kawecka-Jaszcz, 2013). Therefore, a clear definition of HRQOL can be described as 'how well a person *functions* in their life, and their perceived *wellbeing* in all domains of health including physical, mental, and social domains' (Karimi & Brazier, 2016; Kawecka-Jaszcz, 2013; Opara & Jaracz, 2010). An individual's ability to carry out pre-defined activities can be referred to as *functioning* and an individual's subjective feelings refers to their *wellbeing* (Karimi & Brazier, 2016). Therefore, HRQOL has gained special significance in reference to the long-term medical care of chronic illness, as individuals are living longer with supportive medical care (this is discussed in detail in the treatment section) (Kawecka-Jaszcz, 2013).

HRQOL encompass an individual's perception of the impact of health and illness on the physical, mental and social aspects of their life, and is increasingly recognised as a key

outcome measure of health and rehabilitation services (Kawecka-Jaszcz, 2013; Law et al., 2014). Children with GMDs are at a high risk of potentially impaired HRQOL due to living with a chronic and often progressive illness (Landfeldt et al., 2015; Lue, Chen & Lu, 2016). For example, DMD is associated with substantially impaired HRQOL compared to children of the general population (Landfeldt et al., 2015). However, few studies have examined overall HRQOL outcomes more broadly across different GMDs. GMDs vary in terms of the muscles affected, severity, age of onset, and the nature of progression (Landfeldt et al., 2015; Lovering et al., 2005; Lue et al., 2016). A more holistic approach and a better understanding of QOL outcomes across different conditions may reveal important commonalities to inform interventions and support options. Further, impact studies have tended to focus on physical symptoms in adults living with a GMD (Wei et al., 2015). The behavioural and emotional profiles of children living with a GMD have received very little attention and may be affected by their use of coping strategies (Wei et al., 2015; Lue et al., 2016). The few HRQOL paediatric studies are largely limited to parent-report and examine children recruited from single clinics with DMD which limits the extent to which findings can be extrapolated to children living with other GMDs (Landfeldt et al., 2015; Lue et al., 2016; Wei et al., 2015).

Skyrme (2017) and Morris (1992) suggested that children with GMDs are physically limited by pain, discomfort, increased muscle weakness, and a lack of energy. Children living with a GMD also experience negative psychosocial and cognitive effects (Jones et al., 2018; Lovering et al., 2005). For example, Skyrme (2017) found that alongside such physical symptoms, affected children report increased feelings of isolation, discrimination; thus, highlighting a range of negatively impacted HRQOL concepts. Abbott and Carpenter (2015) suggested there is a negative social persona regarding children with GMDs that has a significant impact on community integration of impacted children. Children with a GMD are often limited in their ability to interact socially with their same-aged peers, regardless of social prejudice due to a range of physical, psychological and social barriers (Abbott & Carpenter, 2015; Skyrme, 2017). As a result, this leads to increased risks for social isolation (Abbott & Carpenter, 2005). Skyrme and colleagues (2017) conducted qualitative semi-structured interviews and found that children with GMDs feel insignificant and different compared to their same-aged peers, particularly in terms of their physical status. Abbott and Carpenter (2015) conducted a study on 37 participants living with a GMD residing in England, to explore their life experiences, including childhood development, school, work, friends and family. They found individuals with DMD often experience lower HRQOL in

functional status (not being able to go outside in winter due to risk of attaining a lung infection that may be fatal), fatigue and physical functioning (ability to participate in full time work). Consistent with Skyrme and colleagues (2017) findings regarding impaired physical status. Abbott and Carpenter (2015) reported that children with a GMD have more limited social interactions, according to reports from their primary caregiver/parents. For this was also consistent with Skyrme and colleagues (2017), who highlighted significant impairments in social functioning due to living with a GMDs. A study by Uzark et al. (2012) assessed HRQOL using the Paediatric Quality of Life Inventory (PedsQL) in 117 children living with DMD (6 to 18 years old), their findings suggested younger individuals (between 8 to 12 years) scored significantly higher in physical functioning compared to adolescent (13 to 18 years old) participants. Uzark et al. (2012) also reported children with DMD scored significantly lower than those for healthy children in physical, psychosocial, emotional, social, and school functioning domains by both parent and self-report. Abbott and Carpenter (2015) also reported that children with a GMD have more limited social interactions, according to reports from their primary caregiver/parents.

As mentioned above an important indicator of children's wellbeing is their self-reported HRQOL. This is becoming increasingly recognised as a key outcome of health and rehabilitation services and outcomes (Bann et al., 2015; Engel et al., 2005; Hunt et al., 2016). A pilot mixed-methods study was conducted by Hunt and colleagues (2016) to assess the effectiveness of using specific measures to assess coping and HRQOL, through semi-structured interviews of primary caregivers and boys (the median age was 15 years) with DMD. Hunt and colleagues (2016) concluded that there was no significant difference identified between parent and the boy's ratings on domains of coping. These findings suggest that further research is required to explore the pain experience of boys and young men with GMDs. However, in terms of HRQOL (assessed using the Youth Quality of Life Scale), findings revealed that young men were generally very positive about the quality of their family life. Hunt and colleagues (2016) found that while overall self-reported QOL was good, poorer QOL in young men and children was associated with more moderate to severe pain on a day-to-day basis, according to a parent report. Hunt and colleagues (2016) also found 67% of young males suffered from pain daily, which was associated with reduced QOL. This was consistent with Engel, Kartin and Jaffe (2005), who suggested that boys and young men with DMD are at increased risk of developing maladaptive coping strategies, which consequently, placed increased stress on individuals and their caregivers.

Hunt and colleagues (2016) also emphasised participants often kept pain complaints within the family. Jones and colleagues (2018) identified an increased potential of impaired HRQOL among children with GMDs as a result of living with a prolonged and progressive illness. Another study found increased duration of illness and greater disease severity was associated with lower perceived HRQOL among individuals with a GMDs (Bann et al., 2015). Given there is no cure for GMDs, treatment is often focused on improving physical functioning and perceived HRQOL (Bann et al., 2015; Hunt et al., 2016).

2.1.5 Diagnosis and Treatment

Risk factors differ across the range of possible GMDs, but some common risk factors have been identified. For example, individuals with a family history of GMDs are at a significantly higher risk of developing muscular disorders (Emery, 2002). Such as, males are more likely to develop DMD compared to females (Emery, 2002). In contrast, there are no significant sex differences in congenital myopathies. The most predominant risk factor for congenital myopathies are blood relatives with the condition or either parent (or both) that may be a carrier of the mutated gene (Cassandrini et al., 2017).

Most inherited muscular disorders are diagnosed based on blood tests and genetic testing. This is often supported by family history and family reporting of the symptoms experienced by an individual (Abbott & Carpenter, 2015). As mentioned before, currently, there are no cures for any of the wide-ranging GMDs. However, children are living longer due to advances in supportive medical care including physical, occupational therapy, speech therapy, medication, and orthopaedic treatments (such as braces and corrective surgery) (Gazzard, 2004; Kohler et al., 2009; Strehle et al., 2009). For example, medication is often used to slow muscle degeneration through use of steroids, control seizures by administering anticonvulsants, delay damage to dying muscle cells through immunosuppressants and antibiotics to fight respiratory infections (Abbott & Carpenter, 2015; Lovering et al., 2005; Strehle et al., 2009). Treatment is most commonly focused on symptom management, to optimise QOL and functioning (Huml, 2015). Such as infants with increased muscle weakness or low muscle tone are also likely to benefit from a program involving low impact, mild-to-moderate exercise and stretching techniques (National Organization for Rare Disorders [NORD], 2015).

Treatment is often tailored to an individual's needs or the severity of their symptoms, for example, in severe cases, a pacemaker may be utilised for an individual with cardiac abnormalities or assisted ventilation may be require to treat respiratory muscle weakness; musculoskeletal abnormalities are often treated with use of special orthopaedic braces, other devices, or surgical measures (Huml, 2015; NORD, 2015). The progression of GMDs can vary extensively, while some disorders may progress slowly over a normal lifespan, whereas, others can produce severe muscle weakness, functional disability, and loss of motor skill. In unfortunate cases, some children with GMDs do not make it past infancy while others live into adulthood with only moderate disability (Kohler et al., 2009; NINDS, 2019; NORD, 2015). Family-cantered, early intervention services are often effective, such as physical and occupational therapy, this may also include assistance with seating and mobility devices; educating parents on correct manual handling, exercising and stretching techniques; and other appropriate measures for the specific individual (Kuo, Bird & Tilford, 2011).

2.2 Coping strategies

Together, the challenges of living with a GMD may lead to additional stressors for affected children and their families/whanau as they navigate their daily lives. This situation highlights the potential value of children using effective coping strategies to manage the stressors associated within the context of a progressive GMD. Stress can be defined as a physical, mental, or emotional factor that causes bodily or mental tension. Stressors can be caused by internal or external factors (Krohnea, 2002; Skinner & Zimmer-Gembeck, 2007). It can be viewed as either a response (a physiological response pattern), stimulus (a significant life event that requires a response, adjustment, or adaptation), and/or transaction (stress as a product of a transaction between a person, including multiple systems and his or her complex environment) (Krohnea, 2002; Power, 2004).

Lazarus's Theory of Cognitive appraisal suggests that there must be an appraisal during a time of stress (Krohnea, 2002). For example, an individual will evaluate the significance of what is happening in relation to their wellbeing. This may be followed by consideration of ways to respond to the specific demands of the stressful encounter (Krohnea, 2002; Power, 2004). How an individual conceptualises stress determines his or her response,

adaptation, or coping strategies (Power, 2004). Coping can be defined as "…the cognitive and behavioural efforts made to master, tolerate, or reduce external and internal demands and conflicts among them" (Folkman and Lazarus, 1980, p. 223). The concept of coping assumes the existence of a condition causing stress or adversity (Carver, 2013). Therefore, an individual must deal with the adversity by engaging in coping. Individuals can respond to the perceptions of threat, harm, and loss in a range of ways, many of which are identified as coping strategies. The anticipated fear of an event to have negative consequences can be referred to as a *threat. Harm* can refer to the perception that negative consequences have passed already. *Loss* can often suggest the perception that something of great value has been taken (Carver, 2013). While Compas and colleagues (2001) preferred to limit the concept of coping to only voluntary responses, Skinner and Zimmer-Gembeck (2007) suggest include automatic and involuntary responses. However, it can often be difficult to distinguish between voluntary and involuntary responses to stress. Furthermore, Carver, (2013) suggested that responses that are intentional initially may become automatic with repetition (Carver, 2013; Skinner & Zimmer-Gembeck, 2007). For example, individuals who often 'shutdown' when faced with a difficult situation, are more likely to do it again if it was effective the first time.

Coping can be adaptive or maladaptive. Adaptive coping strategies improve an individuals functioning. Alternatively, maladaptive coping strategies may initially reduce the impact of the stressor but not directly deal with the stressor and may instead reinforce the negative impact of the stressor (Carver & Connor-Smith, 2010). Adaptive coping strategies generally involve problem-solving, positive emotion regulation, cognitive restructuring, seeking social support. Whereas, maladaptive coping strategies often includes blaming others, avoidance, distraction, social withdrawal, wishful thinking, self-criticism, and blaming others. For example, wishful thinking may be beneficial initially but may result in increased social isolation. The current study focuses on adaptive and maladaptive coping stratergies. However another distinction is dispositional and situational coping. Dispositional coping refers to habitual ways of dealing with stressors and trauma that are stable personality traits that developed early in life and are influencing responses across different stressful situations (Carver & Scheier, 1994). Alternatively, coping responses can change from moment to moment depending on the nature and personal appraisal of a stressful transaction, indicating situational coping (Folkman & Lazarus, 1985)

In summary, coping is commonly referred to an individual's intentional efforts to minimise the psychological, physical, or social harm of a stressor, event or circumstance (Carroll, 2013). Coping is considered a very board concept with a complex history (Stanisławski, 2019). A complete review of the development coping strategies can be found in Skinner and colleagues (2003) review and critique of coping. Currently available literature focuses on presenting coping strategies, different frameworks for understanding coping and several ways of classifying types of coping strategies (Carroll, 2013; Stanisławski, 2019; Skinner et al., 2003).

2.2.1 Problem and emotional focused coping

In 1984, Lazarus and Folkman distinguished the two pioneering coping categories, problem and emotion focused coping (Carroll, 2013; Stanisławski, 2019). Problem-focused coping strategies aim to reduce or resolve specific threats or negative consequences (Carver, 2013). Examples of problem-focused coping include problem solving and removing the source of the stress. In contrast, emotion-focused coping strategies focus on the management, minimisation or regulation of emotional distress caused by illness, experiences or perception (Carver, 2013). Examples of emotion-focused coping strategies include distraction, cognitive reappraisal and suppression of undesirable thoughts and feelings (Krohnea, 2002). Both problem and emotional focused coping have different focal and initial goals, however both can be used in parallel with each other. The focal goal usually determines which category a particular response is assigned to. While some behaviours can serve both emotion and problem-focused functioning, this is usually dependant on the underlying goal for their use (Carroll, 2013; Stanisławski, 2019). For example, while using social support, if the goal is to attain advice or help it is considered problem-focused; and if the goal is to gain emotional support and reassurance it can then be emotion-focused coping (Carver, 2013).

Lazarus and Folkman suggested problem-focused strategies are aimed at "managing or altering the problem causing the distress" while emotion-focused strategies are used for "regulating emotional responses to the problem" (Lazarus & Folkman, 1984, p.150). The work of Lazarus and Folkman led the development of the Ways of Coping Questionnaire (WCQ) (Folkman & Lazarus, 1988). The WCQ was used to distinguish between emotion and problem-focused coping to stress during a specific period. Problem-focused coping

behaviours are those aimed at reducing or resolving specific threats or negative consequences, such as controlling symptoms or finding ways to overcome illness-related barriers. Emotion-focused coping involves the management or regulation of emotional distress caused by illness experiences. Emotion-focused strategies may include, for example managing fears about treatment by expressing one's feelings to a family member or seeking reassurance from a doctor.

Eight factors or sub-coping strategies were extracted from the WCQ: problem-solving (when purposeful problem-focused efforts are utilised to alter an adverse situation); escape-avoidance (the use of wishful thinking and behavioural efforts to escape or avoid adverse situation); accepting responsibility (acknowledging one's own role in the problem with the aim of trying to make improve); positive reappraisal (focusing on personal growth and creating positive meaning); confrontive coping (aggressive efforts to alter one's situation); distancing (detach oneself and creating a positive outlook); self-controlling (regulating feelings and actions); and seeking social support (seeking informational and emotional support) (Stanisławski, 2019). Problem-solving, accepting responsibility, confrontive coping are considered as problem-focused coping, whereas, escape-avoidance, positive reappraisal, self-controlling behaviours are considered emotion-focused stratergies.

Despite the impact of Lazarus and Folkman's foundational work, their distinction between problem and emotional focused coping has been criticised for being too simplistic. Skinner and colleagues (2003) suggested that most forms of coping can serve as both emotional and problem-focused coping, and, therefore, can fit into both categories. For example, formulating a plan in response to a stressful situation can guide problem solving processes and also have an emotional impact by calming one's emotional reaction (Stanisławski, 2019). Furthermore, the distinction is not comprehensive as it does not consider other strategies that could be seen as either problem- or emotional-focused strategies (or both), such as seeking social support (Skinner et al., 2003). In 1989, Carver and colleagues build further on Lazarus's model, based on the assumption of emotion and problem focused coping, as they felt it was incomplete. They developed a multidimensional coping inventory, known as COPE, and items with weak loadings were revised or discarded from the final coping strategies. Thirteen coping strategies were identified: five sub-types of problem-focused coping (including active coping, restraint coping, actively seeking social support, planning and suppression of competing activities), five types of emotion-focused

coping were also identified (including social support for emotional support, acceptance, denial, positive grown and turning to religion) and three strategies, that did not fit into either problem or emotional focused were identified as “less useful” strategies. These included focusing on and venting of emotions, behavioural disengagement, and mental disengagement (Stanisławski, 2019).

2.2.2 Engagement and disengagement coping

Carver and Connor-Smith (2010) found the distinction with the greatest importance is ‘engagement vs disengagement’ (Dijkstra & Homan, 2016). Carver and Connor-Smith (2010) suggested coping strategies that aim at dealing with the stressor or stress-related emotions are known as engagement (or approach). In contrast, coping strategies aimed at avoiding confrontation or escaping the threat of stress-related emotions can be identified as disengagement (or avoidance) coping (Moos & Schaefer, 1993; Skinner et al., 2003). Examples of engagement coping strategies can include problem-focused coping (such as problem solving) or emotion-focused coping (such as seeking support, cognitive restructuring and emotion regulation) (Carroll, 2013; Craver, 2013; Dijkstra & Homan, 2016). Disengagement coping strategies include responses such as avoidance, denial, and wishful thinking (Carver & Connor-Smith, 2010). Disengagement coping strategies are often emotion focused. However, these strategies usually involve attempts to escape feelings of distress. For example, an individual may try to act as though the threat does not exist, and therefore requires no reaction emotionally or behaviourally. For example, denial can create a boundary between reality and the individuals experience, another example includes, wishful thinking, which can help distance the individual from the stressor temporarily (Carver & Connor-Smith, 2010).

Dijkstra and Homan (2016) suggested that although disengagement coping strategies aim to escape distress, these strategies are generally ineffective in reducing long-term distress as these do not deal with the threat or its impact. For several types of stressors, the longer a person avoids dealing with the problem, the more difficult or complex it becomes (Carroll, 2013). As a result, leaving individuals less time available to deal with the given problem when it becomes a significant stressor. While other types of disengagement coping can create new problems. Examples include, excessive use of alcohol or drugs can create social and

health issues or using gambling as an escape can often result increased financial pressure (Carroll, 2013; Craver, 2013; Dijkstra and Homan 2016). Previous research centered around coping strategy use have often found disengagement coping strategies are ineffective (Chu-Lien, 2011). This suggest individuals are unable to cope with the stressor and are more likely to experience the negative consequences associated with the specific stressor compared to those individuals who utilise more active coping strategies (Fortes-Ferreira et al., 2006; Chu-Lien, 2011). Dijkstra and Homan (2016) suggested the effectiveness of coping strategies are also driven by the degree to which a person feels a sense of being in control of the stressful situation (Dijkstra & Homan, 2016).

Carver and Connor-Smith, (2010) suggested the concept of disengagement coping can also be extended to include 'giving up on goals' that are compromised by the stressor. 'Giving up on goals' differs from previously mentioned disengagement coping strategies, as it emphasises both the existence of the stressor and the emotional impact it has by abandoning previous set goals. As a result of disengaging from the threatened goal, an individual may avoid negative feels and consequences associated with the stressor or threat. However, this form of disengagement can also have adverse secondary consequences for an individual (Carver & Connor-Smith, 2010). Overall, it is widely accepted that the study of coping is fundamental to furthering our understanding of how stress affects people, in order to establish greater understanding of both effective and ineffective strategies (Carver & Connor-Smith, 2010; Skinner et al., 2003). Although, it has proven difficult to document unequivocally, coping researchers argue how children deal with stress can increase or reduce the effects of adverse life events or conditions. Skinner and colleagues (2003) emphasised this is not only limited to emotional distress and short-term functioning, but also long-term development, such as the development of physical and mental health (Skinner et al., 2003).

2.2.3 Use of coping strategies in children

Spirito, Stark and Williams (1988) suggested there is a lack of literature when considering the use of coping strategies among children and adolescents. This is also true with regards to a lack of literature examining children's use of coping strategies within the context of living with a GMD. Therefore, rather than focusing solely on coping literature among children with

GMDs, the following literature review provides a broad overview of coping strategies in relation to children and adolescents.

It is essential to assess coping in children with serious and chronic illness to provide a better understand of the impact the illness has on the child. In 1999, Pretzlik and Sylva emphasised the importance of coping measures as most paediatric patients are psychologically healthy and, therefore, traditional measures of psychopathology are inadequate in identifying maladaptive behaviours. Distress can be defined as a reaction to stress with emotional implications. As mentioned before, there is variety of definitions available for coping, the root of coping can be defined as cognitive and behavioural efforts to manage and overcome a specific external and/or internal stressor, that is often demanding or exceeding the resources at the disposal of an individual (Peterson, 1989). Distress is therefore seen as an emotional reaction, whereas, coping highlights the efforts an individual utilises to deal with stress (Spirito et al., 1988). Coping strategies used by the individual often varies across types of stressors, the demand exerted from the stressor and over time (Pretzlik & Sylva, 1999).

Better individual functioning in children has been associated with use of active coping strategies, such as problem-solving and monitoring, when compared to less active strategies, such as distraction and self-criticism (Donaldson, Prinstein, Danovsky, & Spirito, 2000). Previous studies of coping in children have examined the frequency of use and effectiveness of coping strategies in relation to a specific stressor. This is often collected as self-reported data by children or their proxy caregiver. Results from previous studies have varied significantly across different stressors (Donaldson et al., 2000). For example, Wertlieb, Wiegel, and Feldstein (1987) reported in response to stressors related to school or with siblings the most frequently used coping strategy was wishful thinking. Whereas, Roecker, Dubow and Donaldson (1996) found children aged between 12 and 14 years reported using distancing or denial more than any other coping strategy.

A Spanish study of 68 internationally adopted children (8 to 12 years old) examined stress, coping and psychological adjustment (Reinoso, Pereda, Van den Dries, & Forero, 2013). Reinoso and colleagues (2013) reported over 65% of children were internationally adopted to Spain from China, Russia and Colombia. Using items on the child self-report Kidcope questionnaire, children were asked to identify the most stressful general and

adoption-related problem they experienced. Children also reported their use and perceived effectiveness of various adaptive and maladaptive coping strategies when dealing with the problem. Findings revealed no differences in terms of coping strategy use in general or in relation to adoption processes, except for 'self-criticism' being used significantly more with general stressful situations (46.8%) compared to adoption-related problems (16.7%). The findings also revealed the importance of identifying the problems and coping strategies of adoptees in order to help these children and their families tackle stressors. This is evident by Reinoso and colleagues (2013) findings that the most commonly reported problems were due to interpersonal relationships. Therefore, although adopted children used a wide variety of coping strategies, which they perceive as highly effective, children may require support with difficulties that may emerge in their interpersonal relationships. Likewise, it is important to consider a large group of adopted children felt victimised when faced with a range of adoption-related stressors. Therefore, Reinoso and colleagues (2013) suggest it is important to consider the implications of feeling victimised when conducting adoptive parent training, developing adoption related policies and integrating children into their new society.

Rodgers and colleagues (2012) examined the use of coping strategies in 40 child cancer patients (7 to 12 years old) who were encountering anticipatory, acute, and delayed chemotherapy-induced nausea and vomiting. Rodgers and colleagues (2012) reported distraction and wishful thinking were the most frequently used coping strategies, whereas, social support and distraction were perceived as the most effective strategies used by children with paediatric cancer. Their study also aimed to identify any differences in coping strategies used by children over time, however, no statistically significant differences were observed. Rodgers and colleagues (2012) also compared the use of active versus passive coping strategies. Their findings suggest that active coping strategies were the most effective for children undergoing chemotherapy. However, Rodgers and colleagues (2012) also concluded that most children did not use active coping strategies. Cognitive or behavioural attempts to directly deal with the problem and their effects can be defined as active coping strategies (such as distraction, emotional regulation, problem solving, cognitive restructuring, and social support). In contrast, passive coping refers to cognitive attempts to avoid actively confronting the problem or behaviours to indirectly reduce emotional tension (such as wishful thinking, social withdrawal, resignation, self-criticism, and blaming others) (Choi, Hegel, Marinucci, Sirrianni, & Bruce, 2011). A recommendation from Rodgers and

colleagues (2012) study suggested children be encouraged to use more active coping strategies while coping with a range of short- and long-term health conditions.

In another Spanish study conducted by Pereda, Forns, Kirchner and Muñoz (2009), the Kidcope questionnaire was used to identify school-age children's stressors and coping strategies in a socio-economically diverse sample. Pereda and colleagues (2009) sought to examine common stressors in children aged between 7 and 12 years from a medium socio-economic status primary school compared to a social child centre from a low socio-economic status neighbourhood. Their findings suggested children considered social support, emotional regulation, and wishful thinking as the most effective coping strategies. In addition, Pereda and colleagues (2009) also found children from lower socio-economic status backgrounds reported increased problems related to 'victimisation and violence,' 'moving to a new house' and conflicts with 'norms and rules.' Children from lower socio-economic status commonly used avoidant strategies compared to children from a medium socio-economic background. This could be related to the perception of not being in control of previously mentioned stressors in lower socio-economic status children (Pereda et al., 2009). Another study found that siblings of young people with a GMD often found it helpful to focus on life one day at a time, restrain negative emotions proactively, and often use humour, and distraction as coping strategies to deal with everyday life stressors (Read, Kinali, Munton, Weaver & Garralda, 2011).

Another study by Smith and colleagues (2013) suggested it is best to view coping as a fluid process for participants. Smith and colleagues (2013) explored the relationship between coping strategies and psychosocial outcomes in adolescents with SCI. SCI can be associated with significant daily challenges and long-lasting impairments in physical, emotional and social domains of health, which can vary with increased injury duration. Smith and colleagues (2013) found coping strategy use and effectiveness was linked with increased impairment, this was consistent with previous literature, which have identified changes in coping strategy use over time among children (Smith et al., 2013). This was in contrast to Pereda and colleagues (2009) who found no difference over time when children were confronted with general stressors. Coping patterns have also been associated with injury severity. For example, SCI with less physical impairment were more likely to blame others. In conclusion, Smith and colleagues (2013) reported that adolescents participants frequently used more cognitive-oriented coping strategies, such as cognitive restructuring and

resignation. This pattern of findings was consistent with adolescents in the general population. Children in the general population often used cognitive-oriented strategies more frequently, however perceived social support and emotional regulation strategies as more effective (Rodgers et al., 2012; Smith et al., 2013). This was also consistent with Rodgers and colleagues (2012) and Bingen et al. (2012) findings. Bingen et al., (2012) also reported social support was identified as the most effective coping strategy (52.6% of those using social support reported it as helping "a lot") at baseline in the paediatric hematopoietic stem cell transplantation (HSCT) sample population. In contrast, although wishful thinking was found to be the most popular (90.0%), it was among the least efficacious (43.2% of those using wishful thinking reported it as "not at all" helpful). There was no significant association between participant demographic or clinical variables and coping strategies used (Bingen et al., 2012).

2.3 Health model: A bio-behavioural model

Relatively little research has examined associations between the types of coping strategies used by children and their self-reported HRQOL. Such research will ideally be informed by a relevant theoretical approach. For example, a bio-behavioural model of paediatric pain has been recommended, in which the use of coping strategies are a crucial intervening factor in predicting the variability in children's' pain perception, pain behaviour, and functional status (Hunt, Carter, Abbott, Parker, Spinty and deGoede, 2016; Krohnea, 2002; Power, 2004).

A Bio-Behavioural model can be applied to a range of health conditions and related outcomes. Such models have been used to examine children's wellbeing across a range of health conditions, including asthma and attention-deficit/hyperactivity disorder (ADHD) (Ghriwati, Winter, Everhart & Fiese, 2017; Vetter et al., 2013). For example, a child's family environment indirectly affected lung function, or with differential roles and impact of anxiety and depression in ADHD (Ghriwati et al., 2017). Varni and Wallander (1988) developed a paediatric stress and coping conceptual model to explain the demonstrated variability in children's adaptation to paediatric chronic physical disorders (Varni et al., 1996) (see figure 1). Pain in paediatric patients is often presented as a cognitive-developmental phenomenon, involving several bio-behavioural components that interact to produce different levels of pain perception and verbal and nonverbal manifestations (Hunt et al., 2016; Vetter, McGwin,

Bridgewater, Madan-Swain & Ascherman, 2013). As a result of this, it is essential to identify other factors that may be influencing pain perception (or other outcomes), and in turn QOL in children. Vetter and colleagues (2013) suggested there is a body of empirical evidence in support of the model. The biopsychosocial model emphasises both the patient's personal perspective but also the parent's own experiential perspective and in turn the interaction and dynamic within the parent-child dyad (Vetter et al., 2013). Therefore, a bio-behavioural model would provide a valuable conceptual framework for the examination of children's coping strategies (and its related factors) within the context of GMDs.

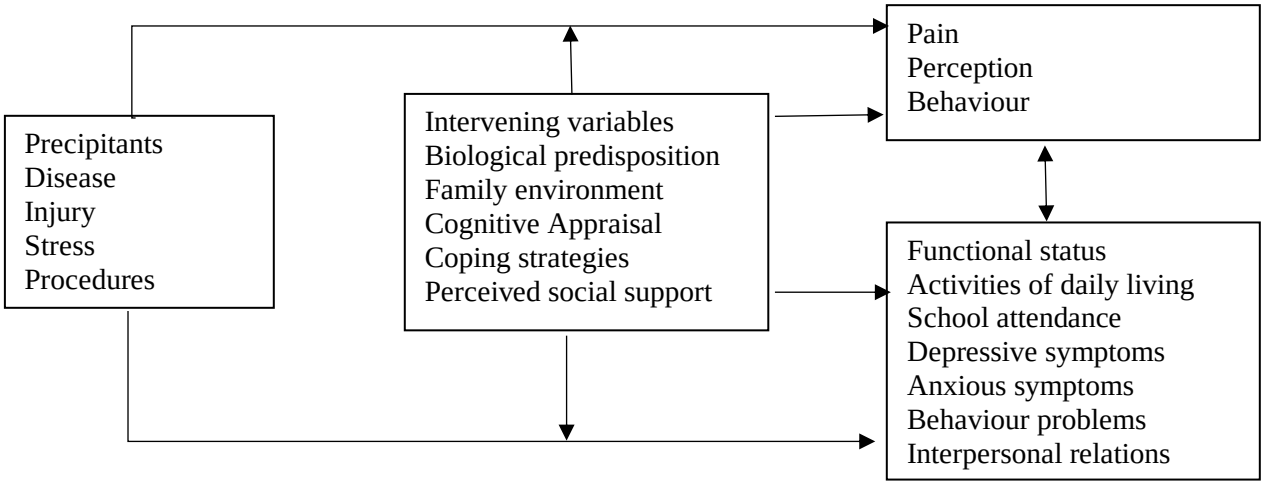

Figure 1. Vetter et al., 2013 biopsychosocial model of health.

In summary, GMDs are a diverse group of hereditary genetic disorders that commonly present with progressive muscle weakness causing significant health impairments and negatively impacting an individual's daily functioning. There is a gap in the literature when considering how children cope with symptoms commonly associated with GMDs and how this may impact children's HRQOL. To date, there is little literature looking at the impact of coping strategies and children's self-reported HRQOL in GMDs populations. The current study aims to understand the use of coping strategies among children with genetic muscular disorders. This Chapter provided a comprehensive introduction to GMDs and coping strategies used by children. The next chapter will outline the methods that were employed to conduct the study.

Chapter Three: Methodology and Method

This thesis is a secondary analysis of pre-existing data collected as part of a nationwide, epidemiological study of the prevalence and impact of GMDs in NZ (known as the MD-PREV study). In brief, the MD-PREV study set out to identify all living adults and children with GMDs, residing in NZ on the 1st of April 2015 (Jones et al., 2018). A complete methodology of the original MD-PREV study has been described in Theadom et al. (2019). This section will provide a brief overview of the main MD-PREV and definition of GMDs an HRQOL. An overview of the current study will also be outlined, including ethical approval, participant inclusion and exclusion criteria, study sample, procedures, measures utilised (predictor and outcome measures), and a brief overview of the data analysis carried out.

3.1 Methodology

The paradigm on which social science and health research disciplines are based on acts like a 'lens', 'filter', or 'framework' through which we see and make sense of research (Halfpenny, 2015). The current study adopts a positivism paradigm, which suggests that real events can be observed empirically and explained through statistical analysis (Sarantakos, 2013). A core strength of using a positivist paradigm is that researchers are required to maintain an external point of view and avoid influencing participant's responses in an effort to minimise researcher bias (Halfpenny, 2015). The role of researchers are often limited to data collection in an effort to remain objective. Data that are objective, observable, and quantifiable may be generalisable to the larger population (DePoy & Gitlin, 2011). Adopting a positivist approach also provides a structure for the current study design, focusing on identifying relationships between variables in specific situations. This paradigm also advocates a 'scientific approach,' that focuses on reporting observable phenomena that can be repeated by others (Dudovskiy 2018).

3.2 MD-PREV Study

The MD-PREV study was funded by the Health Research Council of New Zealand (HRC). MD-PREV aimed to determine age-standardised prevalence of GMDs through a nationwide, epidemiological study of children and adults across the lifespan using the capture-recapture

method on the point prevalence date of April 1, 2015 (Jones et al., 2018). The MD-PREV study also aimed to explore the prevalence of GMDs in NZ by demographic factors, including age, sex, ethnicity, region and disorder type. The study population included individuals of all ages living in NZ (4,242,048), attained though the 2013 NZ population census (NZ Statistics, 2013). The mean age of participants was 39.2 years (SD 20.3) and ranged between 5 months to 90 years of age (Theadom et al., 2019).

In total across all age groups, 966 cases with a confirmed clinical and/or molecular diagnosis of GMDs were identified living in NZ at the point prevalence date (1st April 2015) (Theadom et al., 2019). The crude prevalence was 22.7 per 100,000 person-years (95% CI 21.4–24.3). The mean age at prevalence' is the age of each child on the point prevalence date of April 1, 2015. There were an additional 58 potential cases identified, however, they were excluded as their diagnosis could not be verified. This included 35 self-reported cases, without sufficient clinical and/or molecular diagnosis and 23 individuals who were identified by relatives but did not wish to partake in the study. As there was no way to confirm if they have been identified independently through another case ascertainment source they were ultimately excluded (Theadom et al., 2019).

3.3 Study design

This section will provide insight into the study design. Clarifying the definition of HRQOL and GMDs in context to the current study. The inclusion and exclusion criteria, and participant demographic and GMDs related information.

3.3.1 Ethical approval

Approval for the current study was obtained from the Health and Disability Ethics Committee of New Zealand (Reference number: 14/NTB/118) and the Auckland University of Technology Ethics Committee (Reference number: 14/296) (Appendix 1). All study processes complied with the Helsinki Declaration of 1975. Prior to the collection of any study data, written informed consent was obtained from all parents and assent from children, where deemed appropriate.

3.3.2 Definitions

In order to provide a clear definition of disorders that were included in the MD-PREV study and the current study a definition of GMDs was established based on diagnostic standards outlined by Norwood and colleagues (2009). The term GMDs were defined as inherited disorders that primarily affect the muscles encompassing both non-dystrophic congenital myopathies and muscular dystrophies as well as ion channel muscle diseases (Jones et al., 2018). Disorders of the anterior horn cell, neuromuscular junction and nerves were excluded from the study population (Theadom et al., 2019). An in-depth diagnostic classification can be found in Theadom et al. (2019).

HRQOL can be defined as 'how well an individual function in their life, and their perceived wellbeing across all domains of health including physical, mental, and social domains' (Karimi & Brazier, 2016; Kawecka-Jaszcz, 2013; Opara & Jaracz, 2010). An individual's ability to carry out pre-defined activities can be referred to as functioning and an individual's subjective feelings refers to their wellbeing (Karimi & Brazier, 2016). HRQOL provides a multidimensional approach to examining an individual's wellbeing (Dobkin, 2004; Kawecka-Jaszcz, 2013; Law et al., 2014).

3.3.3 Case ascertainment

Multiple and overlapping sources of case ascertainment were utilised, this included medical record searches tailored to each District Health Board in NZ, using combinations of keywords and/or International Classification of Diseases and Related Health Problems (ICD-10) codes. Similar search strategies were used to check NZ Ministry of Health records, the NZ Neuromuscular Disease Registry, and Genetics Service databases. Advertisements to encourage self-referrals to the study and contact with relevant community support organisations also aided case ascertainment. For all potentially eligible symptomatic and asymptomatic cases, medical records (including investigations and test results) were obtained to confirm details of each diagnosis. Study eligibility was confirmed by a neurologist. Cases with insufficient evidence to confirm a diagnosis were excluded. All cases were screen against the National Health Index number in order to check all new cases against already existing cases, to exclude any duplicates and confirm living status on the point prevalence

date through cross referencing information in the national death registry. Once case ascertainment was completed, participants National Health Index numbers were removed from the main study database. National Health Index numbers were kept in a separate password protected file for the purposes of cross-checking with medical records as required, while protecting patient confidentiality in the main database. A more in-depth case ascertainment is described in Theadom et al. (2019).

3.3.4 Inclusion and exclusion criteria

All confirmed cases of GMDs needed (1) to be either a citizen or resident of NZ (in all child cases, the parent or guardian was required to be registered on the electoral role at a local address or residing in the country for more than 6 months per year); and (2) have a confirmed clinical diagnosis by the child's treating neurologist; and if available, diagnosis were required to be supported by additional lab information, such as, neurophysiological, histochemical, histological and genetic test results. However, generally the treating clinician's diagnosis was accepted. In the case of additional information being available, such as a positive genetic test in a known family member was available, the more specific diagnosis was preferred over the broader diagnosis. If a diagnosis was unclear, a paediatric or adult neurologist reviewed the medical notes and test results. Cases were excluded is there was insufficient evidence to confirm a diagnosis. No additional investigations were undertaken by the research team.

For inclusion in this sub-study, children (aged between 5 and 15 years at the point prevalence date) needed a clinical or molecular confirmation of muscular dystrophy (including Duchenne, Becker, limb-girdle, facioscapulohumeral, Emery-Dreifuss, myotonic dystrophy or congenital muscular dystrophy), congenital myopathy, congenital or juvenile-onset myotonic dystrophy or ion channel muscle diseases (i.e. myotonia congenita or periodic paralysis) (Jones et al., 2018; Theadom et al., 2019). Disorders of the anterior horn cell, neuromuscular junction and nerves were excluded from the study population (Theadom et al., 2019). Generally, metabolic myopathies were also excluded, except where the most usual presentation was of a progressive fixed muscle weakness (such as Pompe's disease) as the dominant feature, this was consistent with previous studies, such as Norwood et al. (2009). While, Duchenne and Becker muscular dystrophy are observed only in males, Female '*manifesting carriers*" were also included in the study to determine the full spectrum of

impact of the condition. As they also experience similar muscular weakness (Theadom et al., 2019). The current study utilised Kidcope and PedsQL data, children who did not complete the measures were excluded from the current study.

3.3.5 Study Sample

The original MD-PREV study identified 159 affected children living in NZ on the point prevalence date. A mean age of 9.47 years (SD 3.92), ranging from 0 to 15 years old (Theadom et al., 2019). The mean age of parent-reported symptom onset being 2.01 years (SD 2.67) and a range of 0 to 11 years, with a large majority of children identifying as male (68.6%) and NZ European (75.9%). Duchenne muscular dystrophy (*n* = 61, 38.3%) and congenital myopathy (*n* = 38, 23.9%) were the most common diagnoses.

Data were available for 133 children living with a GMD in NZ. Data were unavailable for 26 participants. A further, 72 participants were excluded due to various reasons including declining to take part, non-consenting, too busy or too unwell, out of time frame and unknown reasons (figure 2). Due to incomplete and/or missing data for the Kidcope and PedsQL measures, 13 children were removed. Forty-eight children participated in the current secondary analysis, with a mean age of 10.71 (SD 2.89) ranging from 5 to 15 years old (Table 1). A large majority of children identified as male (*n* = 29, 60.40%) and/or NZ European (*n* = 38, 79.2%). Participants were assigned to two age groups, children aged between 5 to 12 years (*n* = 33) and adolescent aged between 13 and 15 years (*n* = 15) at point prevalence. The most common GMDs was DMD (*n* =17, 35.40%).

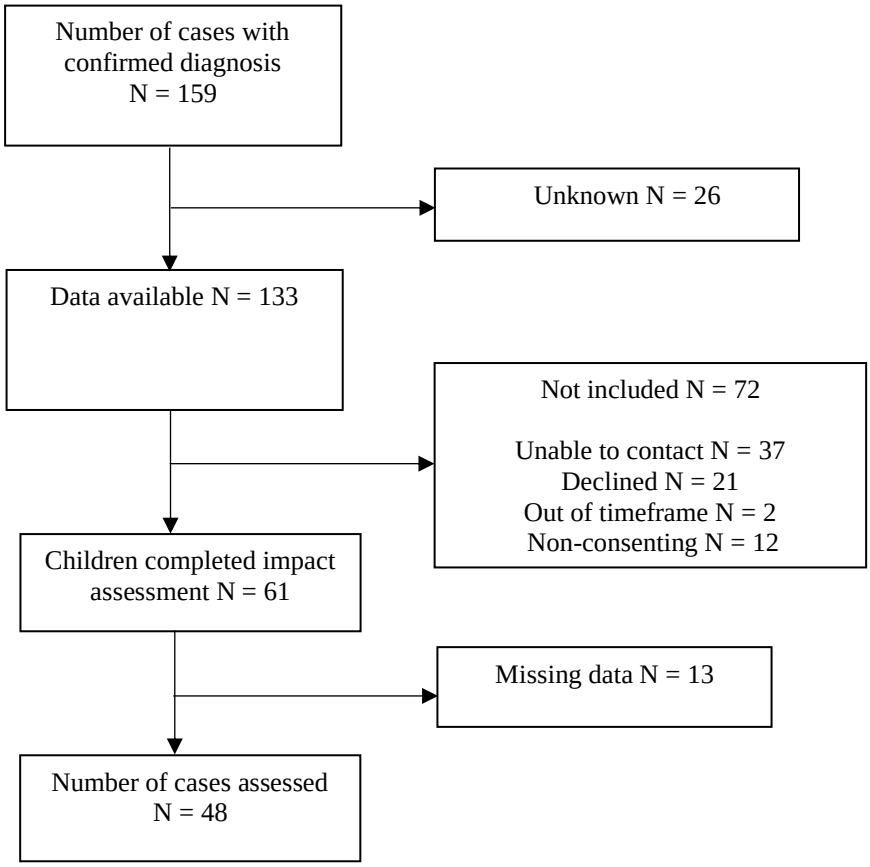

Figure 2. Flowchart of child recruitment.

Profile analysis used descriptive statistics to summarise the demographic, sociodemographic, and GMD related characteristics of participants (Table 1). Reported as means and standard deviations for categorical variables; and number and percentage for continuous variables. To assess the representativeness of the current sample population (N = 48) to available data of children from the original MD-PREV (N = 133). A chi-square analysis was used to compare categorical values, whereas an independent t-test was used to compare continuous variables. A significant difference was observed in the percentage of males included in the current sample compared to the those not included in the analysis (p <.05). there were no other significant differences observed. Therefore, in general the sample population was well representative of the overall population of children living with GMDs in NZ.

Table 1. *Sample characteristics of the current sample (N = 48) compared with participants not included (N = 85)*

Measure	Total sample (N = 133)	Included in analysis (N = 48)	Not included in analysis (N = 85)	t / $\chi 2$	P
Child characteristics					
Mean (SD) age at prevalence	10.81 (2.80)	10.71 (2.89)	11.00 (2.66)	t(131) = 1.66	.56
% Male	95(71.4)	29 (60.4)	66 (77.6)	$\chi 2$ (1, N = 133) = 4.46	.03
Ethnicity					
% NZ European	101 (75.9)	38 (79.2)	63 (74.1)	$\chi 2$ (1, N = 133) = 4.46	.52
% Maori	20 (15.0)	6 (12.5)	14 (16.5)		
% Asian	5 (3.8)	2 (4.2)	3 (3.5)		
% Pacific	6 (4.5)	1 (2.1)	5 (5.9)		
% Other	1 (0.8)	1 (2.1)	0 (0.0)		
GMD diagnosis					
% Duchenne muscular dystrophy	48 (36.1)	17 (35.4)	31 (36.5)	$\chi 2$ (1, N = 133) = .44	.97
% Becker muscular dystrophy	12 (9.0)	4 (8.4)	8 (9.4)		
% Congenital muscular dystrophy	10 (7.6)	4 (8.3)	6 (7.1)		
% Congenital myopathy	22 (16.5)	7 (14.6)	15 (17.6)		
% Other	41 (30.8)	16 (33.3)	25 (29.4)		
% Confirmed genetic diagnosis		29 (60.4)			

Measure	Total sample (N = 133)	Included in analysis (N = 48)	Not included in analysis (N = 85)	t / $\chi 2$	P
% Co-morbid conditions	-	13 (27.1)	-	-	-
% Receiving ventilation support	-	4 (8.3)	-	-	-
% Using a wheelchair	-	27 (56.3)	-	-	-

A dash (-) indicates data not available / not applicable.
‡ Education beyond high school (e.g. Polytechnic, College, University).
* p-value = <.05

3.3.6 Study procedure

As mentioned previously, the current study was a sub-study of the original HRC-funded MD-PREV study. Therefore, permission for the current was formally sought and obtained from the MD-PREV Steering Committee. As part of the original MD-PREV study, parents of affected children were contacted and invited to complete an impact assessment. Written informed consent was obtained from all parents (Appendix B and C). Children were also invited to participate and provided written assent where this was deemed developmentally appropriate. Those consenting to participate were interviewed by a trained researcher either in-person or by telephone.

Children completed the Kidcope – child version (Spirito et al 1988; Pretzlik & Sylva, 1999) which is a 15-item self-report screening tool that assesses the use of 10 different cognitive and behavioural coping strategies. Children were asked to think of a situation when they went to the doctor and weren't very happy about it. Then children rated their use (yes/no) and, if used, efficacy (Not at all, A little or Very much) of each coping strategy. While an adolescent version (for 13+ years) is available, the child version (for 7-12 years) was used by all participants to provide comparable data across younger and older children. HRQoL was assessed using the child (8-12 years) report versions of the 23-item Pediatric Quality of Life with Generic Core Scales (PedsQL GCS) version 4.0 (Varni, Seid & Rode, 1999). The total PedsQL score was used in the current analysis, being an average of the four subdomain scales. Higher scores indicate better HRQOL

Once the study was approved by the MD-PREV Steering Committee, specific data were requested and extracted from the MD-PREV master study database. Kidcope and PedsQL data for all children aged between 5 and 15 years were extracted from the master database. Along with general descriptive information about the sample (i.e. age, gender, ethnicity, type of genetic muscle disorder). Participant demographic information is presented in table 1. All data were kept in password protected documents to ensure confidentiality.

3.4 Measures

This section will provide an overview on the measures used in the current study, including a broad overview on how the measure operates, reliability and validity.

3.4.1 Kidcope self-report questionnaire

As part of the MD-PREV study children completed the Kidcope measure, this was developed in 1988 by Spirito and colleagues. It is a checklist (self-report and proxy parent) created to assess a range of cognitive and behavioural coping strategies. The measure identifies 10 coping strategies. Four of the coping strategies are approach-oriented and are generally considered to be positive or adaptive (problem solving, positive emotion regulation, cognitive restructuring, seeking social support), whereas the other six are considered escape-oriented, and, therefore, generally considered to be maladaptive or negative (distraction, social withdrawal, wishful thinking, self-criticism, blaming others, resignation). The checklist required children and young people to rate the frequency and effectiveness of each of the 10 coping strategies.

The Kidcope checklist consist of four main parts, (1) the stressor within a specific setting, (2) the distress experienced by the child in relation the specific stressor, (3) the child's preferred way of coping with the stressor and (3) the effectiveness of the coping strategy used. Pretzlik and Sylva (1999) suggested it can be repeated across settings and/or over time. The adolescent version of the Kidcope (13 years and older), is comprised of one item per each of the above ten coping categories; whereas, the younger version (for 7 to 12 years) is comprised of 15 items for the same ten scoring categories. Two total scores can be

obtained from the Kidcope measure, firstly the frequency of use (for example, ‘How often did you do this?"), and secondly the effectiveness of the strategy used (for example, "How helpful was it?"). On the younger version, children endorse frequency of the coping strategy as a dichotomous scale (yes and no); in contrast, a five-point Likert scale ranging from not at all (0) to almost all the time (4) is used in the adolescent version. In order to present coping patterns explicitly across both groups of children, frequency items can be converted to dichotomous scores, reflecting whether a coping strategy was used (1) or not (0). On the efficacy scale, younger children respond on a three-point scale (not at all, a little, a lot), whereas adolescent children respond on a five-point scale ranging from "not at all" to "very much" (Spirito et al 1988; Pretzlik & Sylva, 1999).

In terms of reliability and validity of measure, Kidcope results were correlated with results from the Coping Strategies Inventory (CSI) as part of the original development of the measure (Spirito et al., 1988)**.** A moderate correlation was observed between the subscales and trait anxiety when used in a Turkish population, this suggested moderate convergent validity (Bedel, Isik & Hamarta, 2014). Moderate (0.41) to fairly high (0.83) test-retest reliability was observed over a short duration (3 to 7 days) when the measure was originally developed (Sveinbjornsdottir & Thorsteinsson, 2014).

3.4.2 PedsQL

Child self-reported HRQOL was assessed using the 23-item PedsQL with Generic Core Scales version 4.0. The PedsQL assesses four core domains of HRQOL, this include physical functioning (8 items), emotional functioning (5 items), social functioning (5 items), and school functioning (5 items). A psychosocial health score can be generated by averaging emotional, social, and school functioning. A total PedsQL score can also be generated, this is achieved by calculating the average of the four domain (physical, emotional, social and school). Each item is scored using a 5-point Likert scale to indicate difficulties with each item, scores can range from 0 (never) to 4 (almost always) (Varni et al., 1999). Example items included ‘Feeling angry’ (emotional domain) and ‘Paying attention in class’ (school domain). In accordance with standard scoring instructions, each item, including reverse scoring, was rescaled on a 0 to 100 scale (0 = 100, 1=75, 2=50, 3=25 and 4=0). One standard deviation below the mean of the population sample indicates at-risk status for impaired

HRQOL and higher scores indicate better HRQOL (Varni et al., 1999). Along with a total scale score (23 items), physical health scores (8 items) and psychosocial health summary score (15 items) can also be calculated. In order to maximise the availability of data and to select the most robust measure of children's HRQOL, only total HRQOL scores will be examined in the proposed study to allow for a broad understanding of the impact of coping strategies on overall HRQOL. Varni et al. (1999) developed the PedsQL 4.0 Generic Core Scales to be used in a variety of circumstances, including clinical trials, research, clinical practice, school health settings, and community populations.

A study by Viecili and Weiss (2015) assessed HRQOL using the PedsQL in individuals with intellectual and developmental disabilities, such as autism spectrum disorder. Viecili and Weiss (2015) reported high internal consistency across all 4 subscales and the overall total scores. The total score demonstrated excellent internal consistency (alpha = .90) and all subscales had good internal consistency (alpha ranged between .78 - .89). This was consistent with Varni, Seid, and Kurtin (2001) who reported internal consistency was above the minimum of .70 for all self-reported subscales (Varni et al., 2001). Varni and colleagues (2001) also reported high internal consistency and reliability for total PedsQL scores (alpha = .88 child,0.90 parent report), physical health summary score (alpha = .80 child, .88 parent), and psychosocial health summary score (alpha = .83 child, .86 parent). Therefore, concluding it was an acceptable measure to compare within groups. Item-scale correlations demonstrated that most items (19/23) for self-report and all items for proxy-report met or exceeded the .40 standard for item-internal consistency (Varni et al., 2001). Viecili and Weiss (2015) PedsQL had acceptable discriminant, construct, and convergent validity with participants with intellectual/developmental disabilities. Good convergent validity was also observed between PedsQL and Strengths and Difficulties Questionnaire (Viecili & Weiss, 2015).

3.6 Data management and extraction

Data for all study children (5 to 15 years) living with a GMD, who had Kidcope data available were extracted from the master MD-PREV dataset. Data extraction included all subscale and total scores for the Kidcope and child self-reported PedsQL measures. General

participant information on co-morbidities, demographic factors (gender, ethnicity, age at symptom onset, age at prevalence, and ethnicity), diagnosis characteristics, functional status (ACTIVLIM), household income, other family member with weakness, and parent marital status were also extracted in order to provide an overview of the study sample.

All data and participant information will be stored for 16 years in a locked cabinet at AUT University in Auckland by the lead investigator of the MD-PREV study. After 16 years all electronic information will be deleted, and paper forms will be shredded and destroyed with the university confidential waste. Data for the sub-study was managed through a password protected files on AUT's X drive. This was then provided to the primary researcher through a password protected document sent via emailed, followed by a 'naked' email contain the password. Data access was limited to the primary supervisor and researcher. All analysis was run on selected cases (participants), participants without Kidcope data were excluded from all analysis conducted. All missing data were recoded as 999 to avoid it interfering with the analysis.

3.7 Statistical analysis

Logic and data range checks were run to check the completeness of the data. Shapiro–Wilk tests were conducted to determine the distribution of data. Descriptive statistics were used to compare the demographic and clinical characteristics of those children who were included in the current analysis and those children of the same age range who were not included in the current analysis. Comparing the characteristics of these two groups provides an indication of the representativeness of the study sample and the generalisability of study findings to the broader population of children living with a GMDs. This has been presented in table 1.

To address the first aim, coping strategy data were dichotomised to determine whether different types of strategies were used or not (Yes or No). Descriptive statistics were attained for participants that utilised the 10 coping strategies, this was recorded as a percentage. The 10 coping strategies were then grouped into positive and negative coping strategies (as mentioned above) to provide distinct information about the use of adaptive, maladaptive and total coping strategies in the study sample. This was recorded as a median (Md) and interquartile (IR). Children's perceived effectiveness of each type of coping strategy used

was examined using categorical data, this was presented in a bar graph format. Given the number of children that rated the effectiveness of each coping strategy was small (N<34), these findings are reported as percentages to allow for comparisons to be made across the range of coping strategies examined. The perceived effectiveness of positive coping strategies versus negative coping strategies were analysed, firstly a chi-square test of independence was carried out to identify whether there is a link between strategy type and effectiveness. Followed by an independent sample t-test to compare effectiveness between the two strategies (positive and negative).

For Aim 2, chi-square analyses were used to examine associations between each of the 10 Kidcope coping strategies and a range of child demographic factors. Children's use of different types of coping strategies were compared across age (younger [5 years to 12 year] versus older [13 years to 15 years]), sex (male versus female) and ethnic (NZ European, versus Other) groups. A chi-square is a nonparametric test conducted to determine whether there is an association between two categorical variables, such as the use of a specific coping strategy (yes or no) and sex (male or female) (Pallant, 2016). Given the sample size was small, a $p<0.05$ was applied to identify any significant associations between children's use of a specific coping strategy and the above-mentioned demographic factors. A line graph will be utilised to best show the percentage of coping strategy used in each demographic group. The use of positive, negative and total coping strategies will also be presented to identify any differences due to demographic factors. This will be reported as a median and interquartile range. Children were able to select the use of multiple strategies for a single stressor.

HRQOL and coping strategy use (Aim 3) was examined by conducting a Mann-Whitney *U* test. A Mann-Whitney *U* test is a non-parametric alternative to an independent *t*-test (Leech, Barrett & Morgan, 2014; Pallant, 2016). All assumptions of Mann-Whitney *U* were met, including random samples of the population, independence of the data (for example, scores from one participant are not dependent on scores of the others), and the measure of the two samples have at least an ordinal scale of measurement (Brace, Kemp & Sneglar, 2012).). Shapiro-Wilk test was conducted to identify normality of the data, $p<0.05$ indicated the data significantly deviate from a normal distribution (Pallant, 2016). It allows for comparisons between medians of the two groups instead of comparing means of the two groups (t-test) (Pallant, 2016). A Mann-Whitney *U* test will be used to test difference between two independent groups (use of coping strategies, *Yes or No*) on a continuous

measure (HRQOL, PedsQL). The median, *U* statistic, *p*-value and r value will be reported in the results.

As part of exploring any other association between HRQOL and coping strategy use, a Kruskal-Wallis *H* analysis was run to examine any associations between children's use of positive and negative coping strategies and their self-reported HRQOL. A Kruskal-Wallis *H* test is a rank-based non-parametric alternative to a one-way ANOVA analysis (Pallant, 2016). It allows the comparison of scores on continuous variables across three or more groups to determine the presence of any statistically significant group differences (Brace et al.,2012; Pallant, 2016). This analysis was deemed appropriate given there were more than two groups in this part of the analysis, due to the various number of coping strategies used by participants. All assumptions of a Kruskal-Wallis *H* analysis were met, including the dependent variable being measured on a continuous scale (PedsQL) and the independent variable consisting of two or more categorical, independent groups (number of strategies used).

Next, informed by the findings of the Kruskal-Wallis *H* and Mann-Whitney *U* analyses, a hierarchical multiple linear regression was run. Potential issues related to multicollinearity were considered through an inspection of correlation coefficients prior to undertaking regression analyses. Independent variables were entered into the model in two blocks to predict the variance it may explain in total HRQOL score (PedsQL). Following statistical control for child age, sex, and ethnicity, independent variables identified as being associated with the dependant variable was assessed in terms of what it added to the predictive model. Several analyses between the models were run to identify variables that accounted for the largest amount of variance explained. Block 1 examined the amount of variance in self-reported total HRQOL scores that could be explained by child age, sex, and/or ethnicity. Block 2 examined whether children's overall use of positive or negative types of coping strategies accounted for any additional variance in children's HRQOL total scores, over and above any contributions by demographic factors. All analyses were undertaken using the Statistical Package for the Social Sciences (SPSS version 22). Unless otherwise stated, statistically significant was set to $p<0.05$.

Chapter Four: Results

This chapter reports the study findings for each of the study aims. Tables and figures are presented with a brief overview of the key findings.

4.1 Aim 1: Use of coping strategies and effectiveness.

This section will focus on the types of coping strategies used by children living with GMDs and the effectiveness of the coping strategies that were frequently used.

4.1.1 Use of coping strategies

Table 2 reports self-reported use of coping strategies by children and adolescents living with a GMD in NZ. The most commonly used positive coping strategies were cognitive restructuring (*n* = 34) and social support (*n* = 33). Problem solving was the least commonly self-reported positive strategy (*n* = 23) to be used by the sample population. Whereas, the most commonly self-reported negative coping strategy used was wishful thinking (*n* =34), and distraction (*n* = 31). Self-criticism (*n* = 6) was the least used coping strategy, only 12.50% of the total sample population (N = 48) reported using it. There were no significant differences observed across the demographic factors and use of specific coping strategies in the current sample population (this will be present in the next section as part of aim 2).

Table 2 reports median use of positive, negative and total coping strategies across the study sample. The median self-reported use of coping Total Positive Strategies Used (problem solving, emotion regulation, cognitive restructuring, social support) was 3 (IR 1.00), compared to a median of 2 (IR 1.00) for Total Negative Strategies Used (distraction, social withdrawal, wishful thinking, self-criticism, blaming others, resignation). The overall median for total coping strategies used (including both positive and negative strategies) was 5 (*IR* 2.00).

Table 2. *Use of individual and total positive and negative coping strategies by total sample, age, sex, and ethnicity groups.*

Coping Strategies	Total Sample 5-15 years (N = 48)	Age			Sex			Ethnicity		
		Child (N = 33)	Adolescent (N = 15)	*P*	Male (N = 29)	Female (N = 19)	*P*	NZ Euro (N = 38)	Other (N = 10)	*P*
Use of positive coping strategies, n (%)										
Cognitive Restructuring	34 (70.80)	23 (74.19)	11 (73.33)	.95	20 (74.07)	14 (73.68)	.97	27 (75.00)	7 (70.00)	.75
Problem-Solving	23 (47.92)	14 (48.27)	9 (60.00)	.46	16 (59.25)	7 (41.17)	.24	19 (52.77)	4 (50.00)	.88
Emotional Regulation	29 (60.42)	21 (67.74)	8 (55.33)	.27	18 (66.66)	11 (61.11)	.70	23 (65.71)	6 (60.00)	.73
Social support	33 (68.75)	22 (66.67)	11 (73.33)	.64	21 (72.41)	12 (63.15)	.49	25 (65.78)	8 (80.00)	.38
Use of negative coping strategies, n (%)										
Distraction	31 (64.58)	20 (64.51)	11 (73.33)	.55	18 (64.28)	13 (72.22)	.58	25 (67.56)	6 (66.67)	.96
Social Withdrawal	18 (37.50)	9 (30.00)	9 (60.00)	.05	10 (35.71)	8 (47.05)	.45	14 (38.89)	4 (44.44)	.76
Self-criticism	6 (12.50)	4 (12.50)	2 (13.33)	.93	3 (10.71)	3 (15.78)	.60	3 (8.10)	3 (30.00)	.06
Blaming Others	9 (18.75)	7 (21.21)	2 (13.33)	.51	6 (20.68)	3 (15.78)	.67	6 (13.15)	4 (40.00)	.05
Wishful Thinking	34 (70.80)	21 (70.00)	13 (86.67)	.23	21 (75.00)	13 (76.47)	.91	26 (72.22)	8 (88.89)	.30
Resignation	9 (18.75)	5 (15.15)	4 (26.67)	.34	7 (24.13)	2 (10.52)	.23	9 (23.68)	0 (0.00)	.08
Total strategies use	Md (*IR)*									
Total Positive Strategies Used	3 (1.00)	3 (1.00)	3 (3.00)	.69	3 (2.00)	2 (1.00)	.41	3 (2.25)	3 (1.00)	.85
Total Negative Strategies Used	2 (1.00)	2 (2.00)	3 (1.00)	**.04***	2 (1.50)	2 (1.00)	.77	2 (2.00)	2 (2.00)	.83
Total Coping Strategies Used	5 (2.00)	5 (3.00)	5 (3.00)	.23	5 (2.00)	5 (2.00)	.74	5 (2.25)	5 (2.00)	.99

Note. * p-value = <.05.Md = median; IR = Interquartile range.

4.1.2 Effectiveness of coping strategies

Social support (*A lot*, 75.00%) was perceived by children to be the most effective coping strategy. All participants that used Social support voted its effectiveness as either *a little* (25.00%) or *a lot* (75.00%). Whereas, Blaming others was identified as the least effective, with 62.50% of participants identified it as not effective at all, and zero participants identified it as effective (*a lot*). In general, children and adolescents used more positive coping strategies and identified it more effective across all four strategies (cognitive restructuring, problem solving, emotional regulation, and social support). Resignation (55.60%), distraction (50.00%) and social withdrawal (34.78%) was identified as the most effective negative coping strategy (Figure 3).

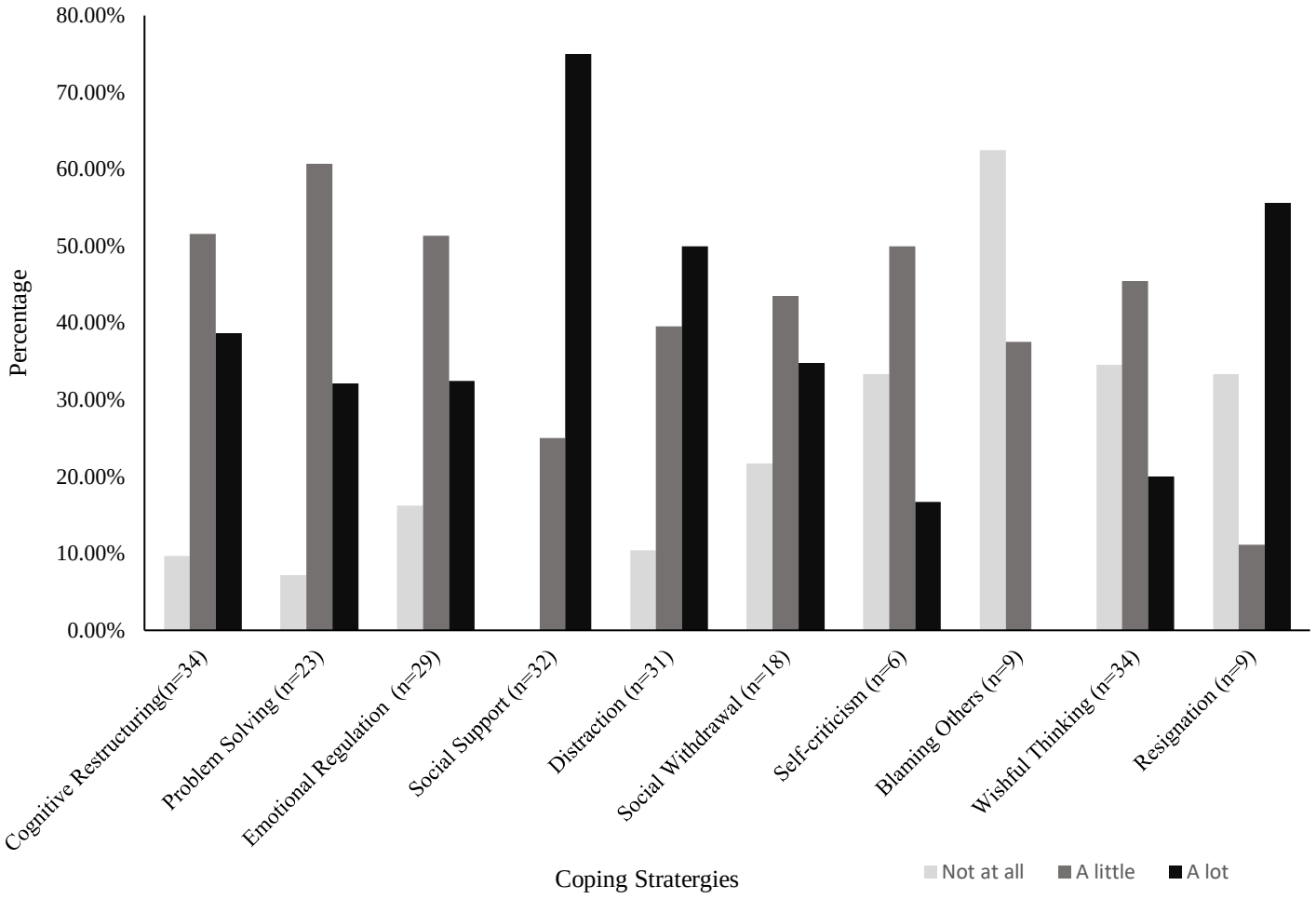

Figure 3. Bar graph highlighting the effectiveness ((Not at all, a little and a lot) of the 10 Kidcope coping strategies used by participants

Table 3 shows the self-reported effectiveness of participants that used positive versus negative coping strategies. A Chi-square test of independence looked at whether there is a link between strategy type and effectiveness. Table 3 shows there was a significant relationship between strategy type and effectiveness, X_2 (*2*) = 14.78, $p < .01$. An independent samples t-test indicated that scores were significantly higher for positive coping strategies (M = 2.36, SD = .64) compared to negative coping strategies (M = 2.07, SD = .77), *t* (275) = 3.41, $p < .001$.

Table 3. *Comparing the perceived effectiveness of positive and negative coping strategies*

Coping Strategy	Mean (SD)	*df*	*t*	Chi square	*P*
		2		14.78	p<0.01*
Total Positive	2.36 (SD .64)	275	3.41		p<0.01*
Total Negative	2.07(SD .77)				

Note. * p-value = <.05

4.2 Aim 2: Use of coping strategies and demographic factors

To understand what extent of children's use of coping strategies differed by their age, sex and/or ethnicity.

4.2.1 Use of coping strategies by age groups

In terms of age, adolescents and children reported using similar types of coping strategies. Table 2 shows a statistically significant difference in medians between use of Total Negative Strategies Used and age groups, Children (5-12 years) median use of negative coping strategies was 2 (IR 2.00), compared to a median of 3 (1.00) for adolescent participants (13-1 years) (p = .04). There were no statically significant differences observed in the individual types of coping strategies nor the overall use of Total Positive Strategies by age groups (p > .05). Figure 4 shows the largest difference in self-reported use of individual coping strategies was observed in the use of social withdrawal (60.00% versus 30.00%, p = .05) between children (5-12 years) and adolescent (13-15 years) participants (Table 2).

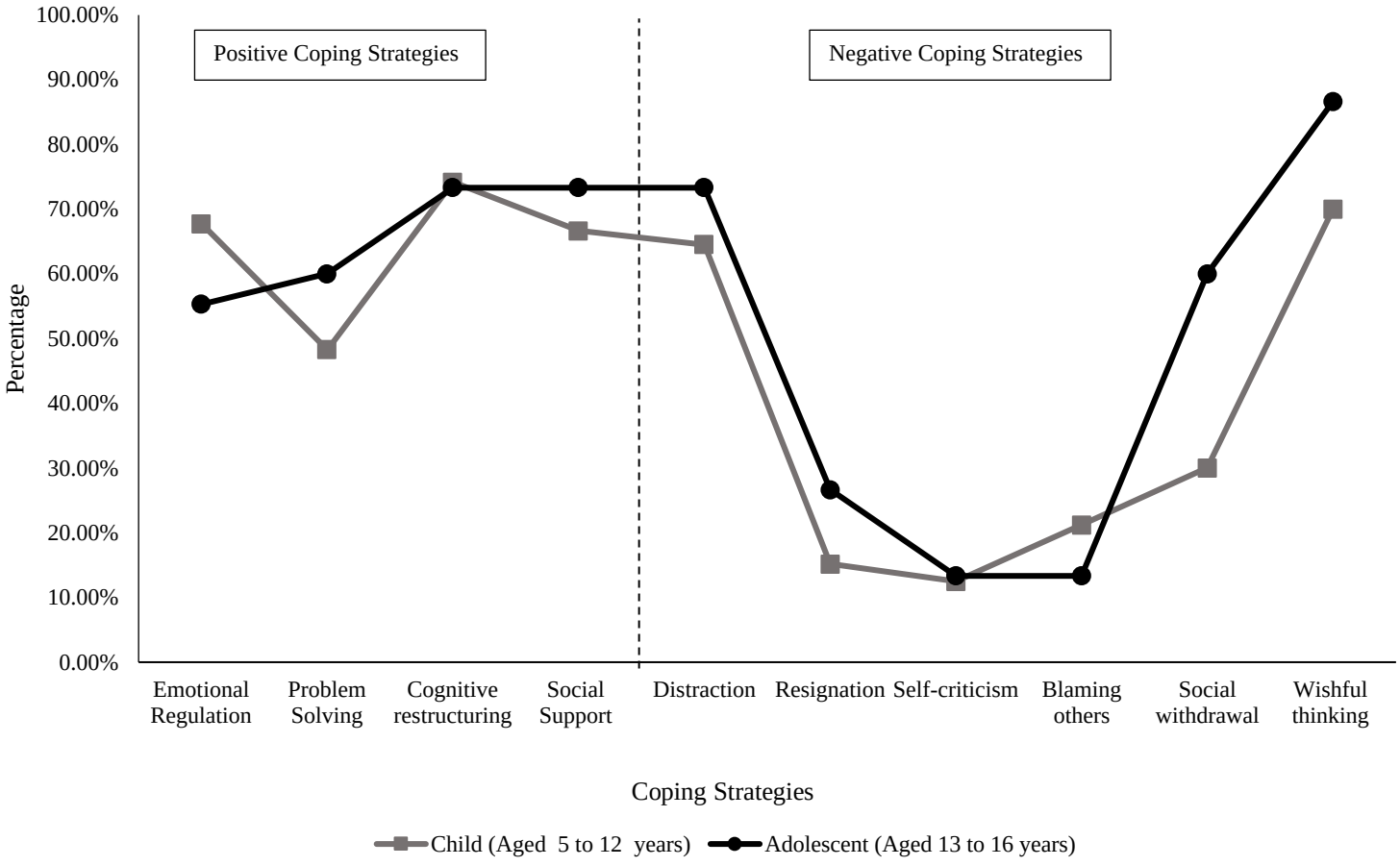

Figure 4. Line graph showing patterns of coping strategy use in children (N = 33) and adolescents (N =15)

In terms of sex, males and females reported using similar types of positive and negative coping strategies (Figure 5). Males (Md= 3, IR =2.00) were more likely to report using positive coping strategies than females (Md= 3, IR =2.00). This pattern of findings was evident across all 4 types of positive coping strategies (problem solving, social support, emotional regulation, cognitive restructuring) as seen on Figure 5. However, group differences by sex did not reach statistical significance (p >.05).

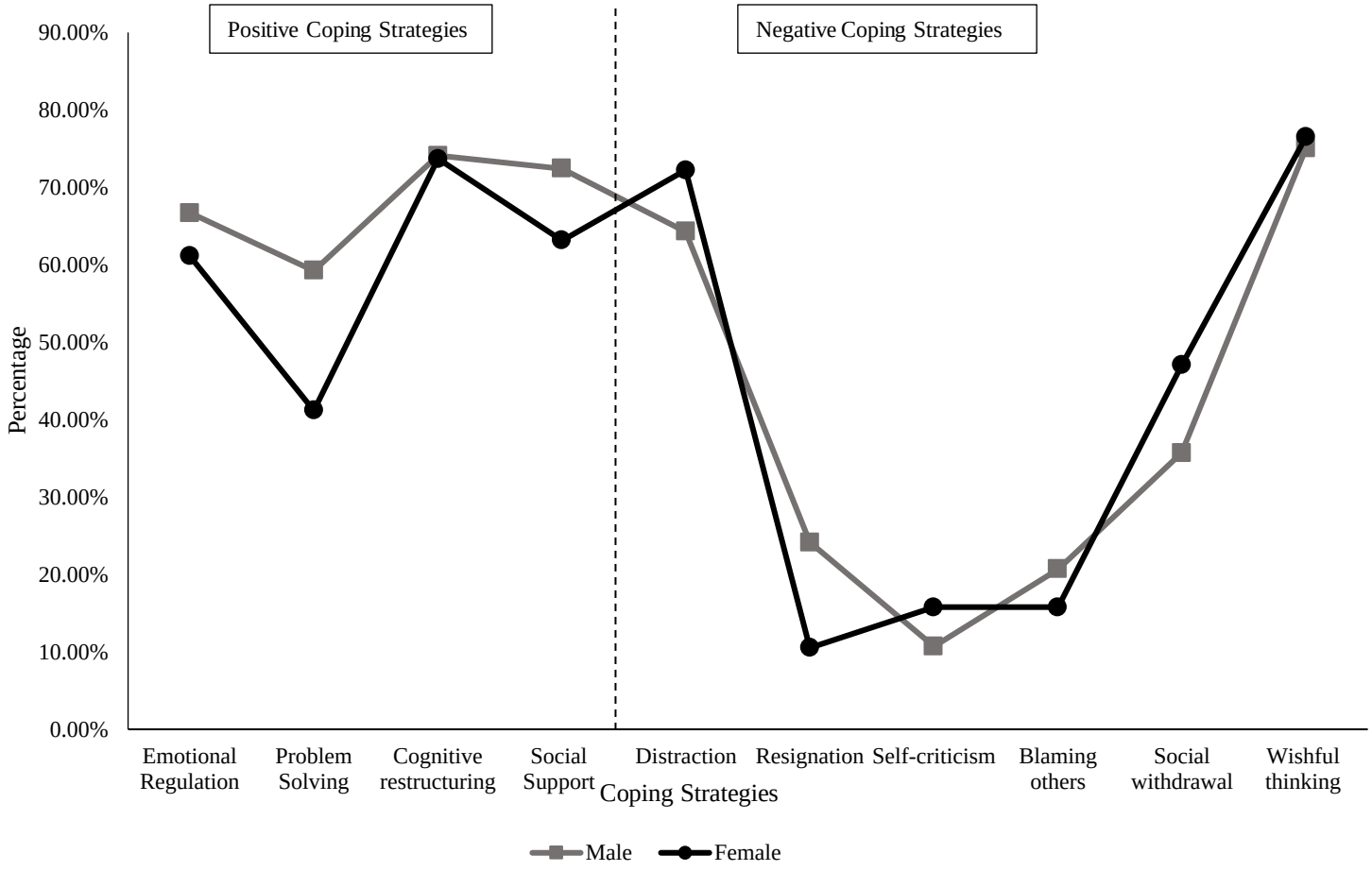

Figure 5. Line graph showing patterns of coping strategies used by male (N =29) and female (N =19) participants in percentages.

4.2.3 Use of coping strategies by ethnicity groups

In terms of ethnicity, children tended to use the same types of positive and negative coping strategies (Figure 6). Compared to NZ Europeans, children in the Other ethnic group reported greater use of negative coping strategies (self-criticism [30.00% vs 8.10%, p = .06] and blaming others [40.00% versus 13.15%, p = .05]). The coping strategy resignation was used by more NZ Europeans compared to Other ethnic groups (23.68% vs 0.00%, *p* = .08). However, these differences across ethnic groups did not reach statistical significance (p >.05).

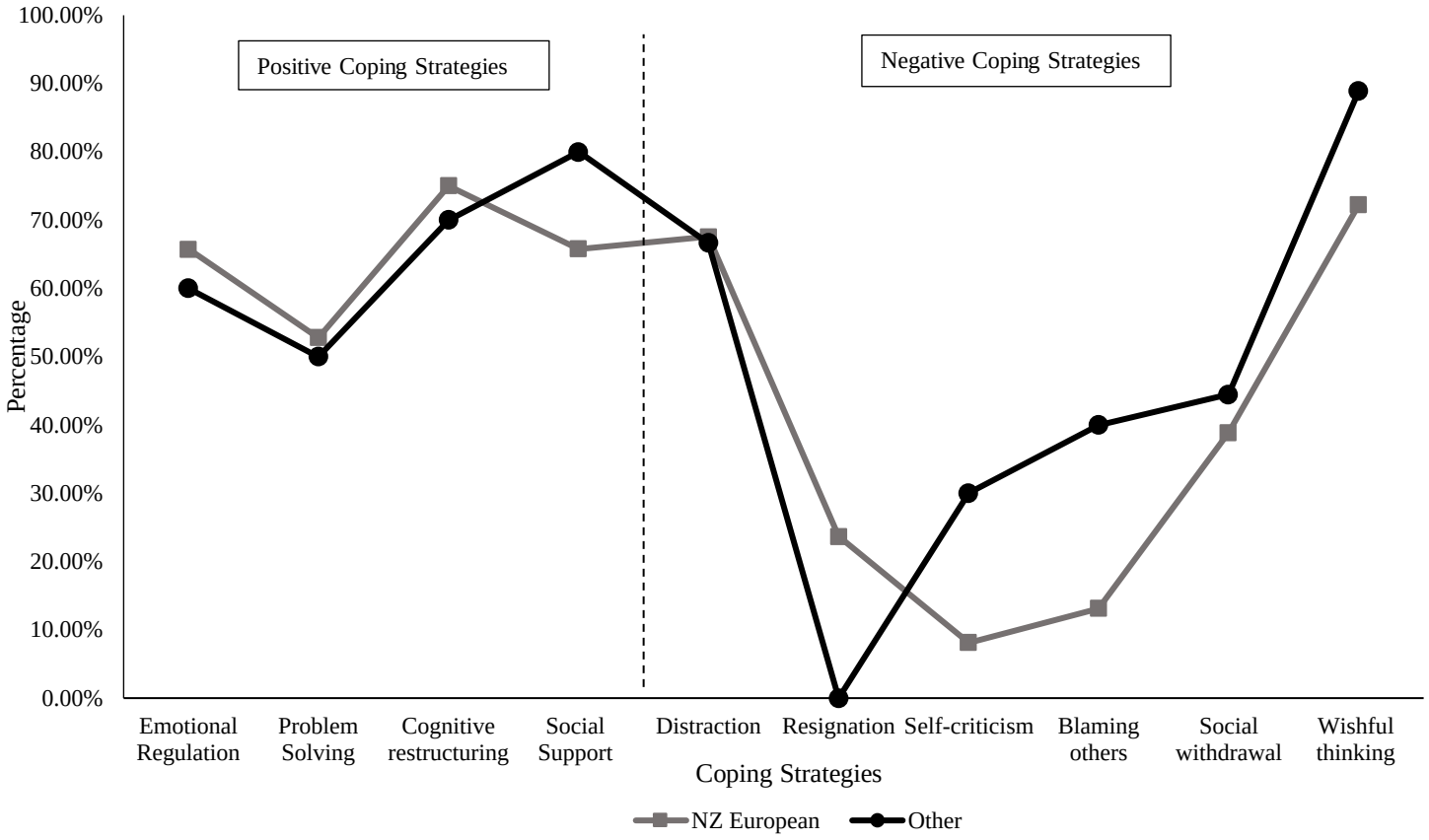

Figure 6. Line graph showing patterns of coping strategies used by different ethnicity, NZ Europeans (N =38) and Other (N =10) participants.

4.3 Aim 3: Associations between coping strategy use and HRQOL

This section will focus on aim 3, reporting relevant findings on the associations between the types of coping strategies used by children and their self-reported HRQOL.

As shown in table 4, a Mann-Whitney *U* analysis revealed significant associations between children's self-reported total HRQOL and two types of positive coping strategies. Those children who reported using problem solving were characterised by lower total HRQOL scores than children who did not report using this strategy (Md = 50.00 versus (vs) Md = 64.13, $U = 138.00$, $z = -2.43$, $p = .02$). Similarly, children who reported using social support reported lower HRQOL than those who did not use this strategy (Md = 50.00 vs 71.74, $U = 88.50$, $z = -3.40$, p = .001).

Mann-Whitney *U* Test also revealed similar associations between children's self-reported HRQOL and two negative types of coping strategies. Those children who reported using wishful thinking were characterised by lower total HRQOL scores than children who did not report using this strategy (Md = 52.17 vs Md = 73.91, $U = 99.00$, $z = -2.33$, $p = .02$). Similarly, children who reported using self-criticism reported lower total HRQOL than those who did not use this strategy (Md = 45.11 vs Md = 59.78, $U = 60.00$, $z = -2.01$, $p = .04$).

Table 4. *Associations between coping strategy use and self-reported total HRQOL in children and adolescents living with a GMD.*

Coping Strategy	Use	N (%)	Median (Md)	Mann-Whitney U (*Z*)	*P*	*R*
Positive coping strategies						
Cognitive Restructuring	Yes	34 (70.8%)	55.98	774.00 (-.63)	0.53	0.93
	No	12 (26.10%)	58.15			
Problem Solving	Yes	23 (47.90%)	50.00	138.00 (-2.43)	0.02*	0.37
	No	21 (43.80%)	64.13			
Social Support	Yes	29 (60.40%)	50.00	88.50 (-3.40)	0.001**	0.51
	No	16 (33.30%)	71.74			
Emotional Regulation	Yes	33 (68.75%)	53.26	166.00 (-1.81)	0.07	0.26
	No	15 (31.25%)	64.1			
Negative coping strategies						
Distraction	Yes	31 (64.60%)	56.52	653.50 (-1.76)	0.07	0.26
	No	15 (31.30%)	64.13			
Social Withdrawal	Yes	18 (37.50%)	50.54	161.00 (-1.90)	0.05	0.28
	No	27 (56.30%)	59.78			
Blaming Others	Yes	9 (18.75%)	41.30	105.50 (-1.85)	0.06	0.27
	No	39 (81.25%)	57.61			
Self-criticism	Yes	6 (12.50%)	45.11	60.00 (-2.01)	0.04*	0.29
	No	41 (85.40%)	59.78			
Wishful Thinking	Yes	34 (70.80%)	52.17	99.00 (-2.33)	0.02*	0.35
	No	11(22.90%)	73.91			
Resignation	Yes	9 (18.75%)	53.26	168.00 (-1.38)	0.17	0.2
	No	39 (81.25%)	56.52			

Note. ** p-value = <.001 * p-value = <.05.

4.3.1 Positive and negative coping strategy use and self-reported HRQOL

Table 5 shows a statistically significant linear relationship between greater use of coping strategies and lower HRQOL, this was significantly correlated for both positive and negative use of coping strategies ($p < 0.01$). A Kruskal-Wallis *H* test showed that there was a statistically significant difference in total HRQOL score between the different number of positive coping strategies used, $\chi_2(4) = 12.69$, $p = 0.01$. Mean rank total HRQOL scores decreased as children used a greater number of positive coping strategies, from 45.38 among those using no strategies to 17.55 among children using four different types of positive coping strategies. Similarly, as mentioned a Kruskal-Wallis H test also revealed a statistically significant linear difference in total HRQOL scores and negative coping strategies used, $\chi_2(4) = 14.19$, $p = 0.01$. Mean rank total HRQOL scores also decreased as children used a greater number of negative coping strategies, from 40.29 among those using no strategies to 18.00 among children using four different types of negative coping strategies.

Table 5. *Total number of positive and negative strategies and median total HRQOL scores.*

Number of Total Strategies	N (%)	Mean Rank	Median PedsQL	Interquartile Range	Chi-Square (*df*)	*p*
Number of positive coping strategies						
Zero strategies	4 (8.30%)	45.38	92.39	21.2	12.69(*4*)	0.01*
One strategy	6 (12.50%)	27.17	59.24	35.33		
Two strategies	12 (25.00%)	26.25	58.70	14.95		
Three strategies	15 (31.30%)	21.57	56.52	30.43		
Four strategies	11 (22.90%)	17.55	48.91	19.57		
Number of negative coping strategies						
Zero strategies	7 (14.60%)	40.29	85.87	31.52	14.19(*4*)	0.01*
One strategy	4 (8.30%)	33.00	73.91	-		
Two strategies	16 (33.30%)	23.09	51.09	22.83		
Three strategies	16 (33.30%)	18.91	46.2	29.35		
Four strategies	5 (10.40%)	18.00	55.43	22.83		

* p-value = <.05. PedsQL = Paediatric Quality of Life Inventory version 4.0, a dash (-) indicates data not available / not applicable.

4.3.2 Predictors of child self-reported HRQOL

Table 6 reports the results of a hierarchical multiple linear regression model examining possible predictors of children's total HRQOL scores. First, key child factors (age, sex, ethnicity) were entered into the model to statistically control for factors commonly associated with children's outcomes (Model 1). Child ethnicity made a significant independent contributions to children's total HRQOL (β=-.33, p = .03). Child age and sex did not make significant independent contributions to children's self-reported HRQOL (p >.05). This initial model accounted for 11% of the variance in children's total HRQOL (p >.05).

In addition to those child factors entered Model 1, the total number of positive and negative coping strategies used by children were added to further extend the model. As shown in Model 2, the addition of children's positive and negative coping strategy use accounted for 49% of variance in total HRQOL scores (p < 0.01). Negative coping strategies with a beta coefficient of -0.44 was the largest coefficient in the model, making the strongest unique contribution to the model (β=-.44, p < .01).

Table 6. *Significant predictors of child self-reported HRQOL in children living with a GMDs*.

	Model 1 (R2 = .11, F= 1.72, p=0.18)			Model 2 (R2 = .49, F= 7.74, p=<0.01)		
	B (SE)	*β*	*P*	B (SE)	*β*	*P*
Demographic factors						
Age	-0.94 (0.99)	-0.14	0.35	0.17 (.80)	0.02	0.83
Sex	2.61 (5.62)	0.07	0.64	2.28 (4.39)	0.06	0.61
Ethnicity	-14.57 (6.67)	-0.33	0.03*	-11.52 (5.20)	-0.26	0.03*
Total coping strategies used						
Positive Coping Strategies				-4.85 (1.87)	-0.32	0.01*
Negative Coping Strategies				-6.76 (1.97)	-0.44	p<.01*

Chapter Five: Discussion/Conclusion

The study sought to explore the types and effectiveness of coping strategies used by children living with a GMD. to highlight any differences in coping strategies used due to demographic factors (age, gender and ethnicity). This study also wanted to determine if there was an association between the coping strategy used and self-reported HRQOL. The key findings of the study highlighted that a range of positive and negative coping strategies were used by children. This was predominatly consistent with previous literature. A significant difference was observed between use of negative coping strategies and children (5-12 years) compared to adolescents (13 to 15 years). However, no significant difference was observed between individual use of coping strategies and demographic factors. A significant association was identified between the use of coping strategies and HRQOL in children with GMDs. This discussion will focus on highlighting the findings of the study in relation to previous literature and provide an insight into the relevance thereof for children living with GMDs. Initially, the use of coping strategies and their effectiveness will be discussed, followed by the impact of demographic factors on the use of coping strategies. Finally, the use of coping strategies will be discussed in relation to the impairments in domains of HRQOL previously observed in the literature, incorporating both GMDs specific literature and general paediatric populations.

5.1 Coping strategies use and effectiveness

In terms of the coping strategies used by children, cognitive restructuring and wishful thinking were most commonly used, followed by social support and then distraction. Cognitive restructuring and wishful thinking may have been the most commonly used strategies; however, children did not perceive them as the most effective strategies. Social support was perceived as the most effective positive coping strategy; resignation and distraction were perceived as the most effective negative coping strategies. In summary, the findings of current study were mostly in accordance with previous literature regarding the use of coping strategies and the effectiveness of strategies among children and adolescents (Bingen et al., 2012; Rodgers et al., 2012; Pereda et al. 2009; Smith et al., 2013). Also using the Kidcope, Smith and colleagues (2013) examined coping strategy use in teenage children (aged 13 to 18 years) with SCI in the United States of America (USA). Smith and colleagues

(2013) concluded that cognitive restructuring and resignation were the most commonly used coping strategies, whereas social support, emotional regulation and cognitive restructuring were perceived as the most effective coping strategies (Smith et al., 2013). Consistent with Smith et al. (2013) findings the current study identified children perceived social support as an effective coping strategy. However, the results of the current study also varied compared to Smith et al. (2013) findings, in that while cognitive restructuring was commonly used in both studies, the current study children did not perceive the strategy as effective in contrast to Smith et al. (2013).

Cognitive restructuring and seeking social support are considered positive forms of coping that are aimed at dealing with the stressor or stress-related emotions (Carroll, 2013; Craver, 2013). In contrast, disengagement or negative types of coping strategies (such as wishful thinking and distraction) aim to escape stress, instead of dealing with the stressor (Dijkstra & Homan, 2016). Cognitive restructuring is the process of learning to identify irrational or maladaptive thoughts (known as cognitive distortions) of the self, world or future (Hope, Burns, Hayes, Herbert, & Warner, 2007). This learning is then applied to rationally dispute negative or maladaptive thoughts and subsequently develop more rational and adaptive thoughts (Hope et al., 2007). This is an important coping strategy to be considered within the context of living with a GMD, as unfortunately, physical disabilities are often permanent, irreversible and increasing over time. In light of findings psychological education around the use of cognitive restructuring may be useful in supporting children with a GMD. Acceptance Commitment Therapy (ACT), for example, may provide a valuable addition to children's 'toolbox' for coping with difficult everyday situations due to living with a GMD. Cognitive restructuring may provide children with a long-term adaptive coping strategy that may be more effective compared to wishful thinking and distraction.

As mentioned earlier, wishful thinking and distraction were the two most commonly used negative coping strategies reported by children in this study. This finding are consistent with Rodgers et al., (2012) who found that children (aged 7-12 years) with cancer were most likely to use distraction and wishful thinking, despite reporting the most effective coping strategies were social support and distraction. Findings of the current study aligned with Rodgers et al., (2012), as distraction was identified as the third most effective coping strategy used by children with a GMD. A possible explanation for the increased effectiveness of distraction as a coping method may be best explained within the context of living with

increasing chronic pain. Skyrme (2017) suggested that children with GMDs are physically limited by pain, discomfort and increased muscle weakness. Therefore, the use of distraction may provide children with a short-term or temporary solution. This may also be the case for other children, such as children suffering from the side effects of chemotherapy-induced nausea and vomiting (Rodgers et al., 2012)

While children in this and previous studies have found distraction to be an effective coping strategy, there may be issues associated with this strategy. Dijkstra and Homan (2016) suggested that distraction and wishful thinking are generally ineffective in reducing distress as these do not deal with the threat or its impact in the long-term. Dijkstra and Homan (2016) conducted a study (N = 543) in the Netherlands, comparing the effectiveness of coping strategies in terms of reducing the negative effect of 'everyday' stressors on wellbeing. Dijkstra and Homan (2016) found active coping strategies were associated with increased sense of control and long-term psychological well-being, in contrast disengagement coping, such as distraction and wishful thinking were associated with less perceived control, and negatively associated with long-term psychological wellbeing. For example, wishful thinking can often distance the person from the stressor, at least temporarily, and distraction can provide the individual with an alternative to the stressor without addressing the stressor (Carver & Connor-Smith, 2010). However, this does not directly deal with the stressor and therefore does not provide a long-term solution (Dijkstra & Homan, 2016). Therefore, wishful thinking may provide children with GMDs with an 'escape' from their disorder. Wishful thinking may provide children with the ability to imagine life as a child without a GMD. However, as previously mentioned, this strategy does not address the long-term impact of living with a GMD. This may explain the finding in this study with regards to wishful thinking being commonly used but not considered to be an effective coping strategy for children living with a GMD in NZ. This pattern of findings are consistent with Rodgers et al. (2012) who found that while wishful thinking was commonly used by paediatric cancer patients, it was perceived as an ineffective coping strategy. In summary, distraction and wishful thinking coping strategies may not be considered effective in the general paediatric population when dealing with everyday stressors. However, when considering children living or experiencing a debilitating health condition distraction and wishful thinking may provide a short-term solution.

The current study found that social support was commonly used by children living with a GMD and was considered to be an effective coping strategy. This is consistent with findings from several previous studies across a range of paediatric populations, including children living with cancer (Rodgers et al. 2012), HSCT (Bingen et al., 2012), SCI (Smith et al., 2013), and a variety of stressors (e.g., school, parents/family, siblings, or peer/interpersonal) (Donaldson et al., 2000; Pereda et al., 2009). Social support refers to informative, emotional or instrumental support received from family and/or friends that may improve an individual's self-esteem or provide stress-related interpersonal help (Kim, Jeong Yeob Han, Shaw, McTavish, & Gustafson, 2010). Social support provides a buffer to stress while providing a valuable resource for coping with stressors (Aflakseir, 2010; Carroll, 2013). For example, children spending time with family or friends may help avoid stressful situations such as social isolation, and the increased social and emotional support may also aid the individuals ability to cope with the stress (Aflakseir, 2010; Rodgers et al., 2012). In the current study, the majority (75%) of children found that social support (e.g. spending time with their family and friends) made them feel better. This finding is consistent with the results of Bingen et al. (2012) study of 93 children (aged 7 to18 years) with HSCT. Results showed that children were most likely to use wishful thinking as a coping strategy; however, wishful thinking was also perceived as the most ineffective strategy used by children. Half (52%) of children in Bingen's (2012) study considered social support as the most effective coping strategy. Similarly, using Kidcope, Pereda et al., (2009) study of Spanish school-aged children living in medium and low socio-economic status neighbourhoods found that one third of children also reported social support as the most effective coping strategy across a range of problems/stressors (e.g., victimisation, violence, moving to a new house, and conflicts with norms and rules). Although Smith and colleagues (2013) found that children were more likely to use cognitive-oriented strategies; social support and emotional regulation strategies were considered by children to be more effective. While children in the current study commonly reported using wishful thinking and emotional regulation coping strategies, these strategies were not perceived to be as effective as social support.

Given social support is consistently acknowledged as being effective across a range of paediatric populations it is crucial to building strong social circles or support channels for children with GMD to provide them with stable and adaptive coping mechanisms. Abbott and Carpenter (2005) suggested that there is a negative social persona regarding children with

GMDs that has a significant impact on community integration of impacted children. Children with a GMD are often limited in their ability to interact socially with their same-aged peers, regardless of social prejudice due to a range of physical, psychological and social barriers (Abbott & Carpenter, 2005; Skyrme, 2017). Therefore, based on the findings of this study and previous literature, supporting children in their capacity to initiate, build and maintain positive social relationships with adults and peers may provide important opportunities to help children effectively cope with living with a GMD.

The findings of the current study indicated children living with GMDs perceived positive coping strategies (cognitive restructuring, problem solving, emotional regulation, and social support) to be significantly more effective than negative coping strategies (distraction, social withdrawal, wishful thinking, self-criticism, blaming others, resignation). Rodgers and colleagues (2012) findings suggest that while active coping strategies (cognitive restructuring, problem solving and emotional regulation) were the most effective for children undergoing chemotherapy, they were not the most commonly used strategies. Whereas, the current study reported that positive coping strategies were used more commonly compared to negative coping strategies and perceived as the most effective. As mentioned earlier, active coping strategies, such as problem-solving, are related to better functioning when compared to less active strategies, such as distraction (Donaldson et al., 2000). This is an encouraging finding of the current study as it suggests that children living with GMDs have some appropriate and accurate level of insight into the value of using positive, rather than negative coping strategies. Previous literature has often identified demographic factors as a significant contributor to the use of coping strategies. Demographic factors and the use of coping strategies in children living with a GMD will be presented next.

5.2 Coping strategy use by demographic factors

In terms of age, the current study found that children and adolescents reported using similar types of coping strategies. This pattern of findings was in contrast to those of previous findings (Donaldson et al., 2000; Spirito et al., 1991; Hampel & Petermann, 2005). Hampel and Petermann's (2005), using the German Coping Questionnaire for Children and Adolescents, reported that emotional regulation and distraction were the more commonly used coping strategies in younger (8 to 10 years) rather than older (11 to 14 years) children

when dealing with situation-specific (interpersonal and an academic) stressors. Hampel and Petermann (2005) findings of age-related differences in coping strategies used were also consistent with Spirito et al. (1991) and Donaldson et al. (2000). Donaldson and colleagues (2000) reported a significant decrease in the use of distraction as a coping strategy from early (9 to 14 years) to middle adolescence (14 to 17 years). Using the Kidcope measure, Donaldson et al. (2000) examined the coping strategies of 599 children across 4 types of key stressors (school, parents/family, siblings, interpersonal). Whereas, Spirito et al. (1991) explore the linear relation between age and use of coping strategies. Spirito and colleagues (1991) reported a significant linear relation between the increase in age of children (9 to 14 years old) and the decreased use of distraction as a coping strategy. Therefore, younger children used distraction as a coping strategy more commonly when compared to older/adolescent children (Spirito et al., 1991). In contrast to previous literature, no significant age-related changes in children's use of distraction and emotional regulation coping strategies were observed in the present study. There are several differences to consider between our study and the Donaldson et al. (2000) and Hampel and Petermann (2005) studies. These include sample size, with both of these studies examining a considerably larger sample size (599 and 1123 participants) compared to the current study (48 participants). Another difference that may have contributed to the differences observed in findings may be due to the varying age groups examined and the stressors children were responding to.

Consistent with the current study, Donaldson et al. (2000) found no statistically significant differences in children's use of wishful thinking and self-criticism by age groups. This finding is consistent with those of the present study where, as no significant age-related differences were observed in the use of either coping strategy. The largest difference in self-reported individual coping strategies across age groups in the current study was observed in the use of social withdrawal between children and adolescents. This association may have been significant if the population size was larger.

In contrast, a significant difference was observed between the use of total negative strategies and age groups. Children's (aged 5 to15 years) were less likely to use negative coping strategies than adolescents (aged 13 to 15 years). This patterns of age-related differences in the use of positive and negative coping strategies is consistent with previous literature, reporting increased use of social withdrawal, distraction and wishful thinking in

adolescent participants (Donaldson et al., 2000). Compared to younger children, Hampel and Petermann (2005) also found that adolescent males and females used more maladaptive strategies compared to adaptive strategies when faced with interpersonal and an academic stressor. The findings of the current study regarding increased use of negative coping strategies in adolescents may be anticipated in children with GMDs. Social withdrawal, distraction and wishful thinking were the most commonly used negative coping strategies by adolescent participants compared to younger children. This can be contributed to the impact GMDs have on participants daily function and consequently their HRQOL. Landfeldt et al. (2015) reported children living with a GMD have substantially impaired HRQOL compared to children of the general population in all domains of health, including social and emotional. As individuals with GMDs are often experiencing psychological impairments along with experiencing progressively worse physical symptoms, including progressive muscle impairment, increased muscle weakness and extreme discomfort which eventually results in severe impairment (Emery 2002; Skyme, 2017). Therefore, contributing to the increased use of negative coping strategies, such as social withdrawal and distraction to avoid the progressive difficulties experienced by children as they age.

With regards to sex, there were no significant differences in coping strategy use among males and females. Males were more likely to use each of the four positive coping strategies than females. This pattern of findings is consistent with Donaldson et al. (2000), who reported no significant difference between sex and use of coping strategies. However, this was in contrast to Hampel and Petermann (2005) who reported that females used more maladaptive coping strategies, such as social withdrawal compared to males. Greater use of negative coping patterns among female children and adolescents compared to males have been reported in several studies (Hampel & Petermann 2005; de Anda et al., 2000). For example, de Anda and colleagues (2000) examination of 333 adolescents (15 to 18-year-olds) in Los Angeles found that females were less likely than males to use emotional regulation and problem solving; and more likely to use resignation and self-criticism (de Anda et al., 2000). As mentioned above, the findings of the current study were not consistent with previous literature. The current sample population consisted of 40% females compared to de Anda et al. (2000) who examined a slightly higher percentage of females (55%). This may have contributed to the difference observed in the previous literature.

In terms of ethnicity, there were no statistically significant associations identified between NZ European and other ethnic group children and coping strategy use. In the current sample population, a large majority of participants identified as NZ European and a small minority were grouped into the other ethnic group (including Maori, Asian, and Pacific) as seen on Table 1. Compared to NZ Europeans, children in the other ethnic group reported greater use of negative coping strategies, including self-criticism, and blaming others. There were no differences observed in the use of social support by ethnicity. There is extremely limited literature regarding the use of copings strategies in NZ (Jose & Schurer, 2009). Jose and Schurer (2009) reported similar use of social support across 556 adolescent NZ Europeans, Maori, and Asian participants, and no significant difference across the three groups when comparing the general use of coping strategies. However, Jose and Schurer (2009) reported Asian and Māori adolescents reported higher levels of problem solving than NZ European participants. This was again inconsistent with the findings of the current study. Jose & Schurer (2009) findings were consistent with the previous body of literature, as Clarke and Jensen (1997) also identified no significant cultural differences in how social support was used by Māori and NZ Europeans. de Anda et al. (2000) compared the use of adaptive coping strategies in European Americans compared to African Americans. Their results indicated European-Americans used more adaptive coping strategies when factors like socioeconomic status were controlled.

Further research is needed to clarify and add to the current findings. In summary, demographic factors were not significantly correlated with use of individual coping strategies. This contrasted with previous literature, there was also limited literature comparing the use and effectiveness of coping strategy use in NZ Europeans and other ethnicities. Thus far, this discussion has briefly highlighted the impact GMDs have on several domains of children's health and emphasised this is not only limited to physical impairment. The last part of this discussion will focus on integrating the use of coping strategies and the influence it may have on children's self-reported HRQOL.

5.3 Association between coping strategy use and HRQOL

In paediatric chronic illnesses, improving HRQOL has become one of the most important goals of disease management (Wei et al., 2015). Coping strategies are an important factor to

consider, in order to provide a multidimensional solution to improve all aspects of functioning in individuals living with GMDs. As mentioned throughout this thesis, there is little research that has examined associations between the types of coping strategies used by children and their self-reported HRQOL. A bio-behavioural model of paediatric pain can be applied across a range of relative health conditions, including GMDs (Vetter et al., 2013). Perception of pain in paediatric patients can be influenced by several components, as identified in the bio-behavioural model (figure 1). Therefore, the use of coping strategies can have an impact on various aspects of an individual's life, including functional status, daily functioning, and increased depression and anxiety. This further highlighted the need to identify coping strategies that could be effective in improving HRQOQL in children. The findings of the current study identified a significant association between the use of social support, problem solving, wishful thinking and self-criticism coping strategies correlated with lower total HRQOL scores in individuals living with GMDs. No significant association was identified in the other six coping strategies studied. The use of effective coping strategies such as social support and cognitive restructuring can improve domains of health as discussed previously (Kim et al., 2010). However, there is a lack of literature exploring the effectiveness of coping strategies and the impact it has on the HRQOL of children living with a GMD (Landfeldt et al., 2015; Lue et al., 2016; Wei et al., 2015). This section of the discussion will draw from a broad range of paediatric literature, integrating HRQOL finings in GMDs and use of coping strategies in a range of health-related conditions.

In the current study, 49% of the variance in HRQOL was explained by ethnicity and the use of negative and positive coping strategies. Therefore, the use of coping strategies was identified as a significant predictor of total HRQOL in children with GMDs. In summary, several previous studies have identified significant impairment of multiple HRQOL domains in GMDs populations across many different cultures (Abbott & Carpenter, 2015; Skyrme, 2017; Uzark et al., 2012). Abbott and Carpenter (2015) conducted a study on 37 participants living with a GMD residing in England, to explore their life experiences, including childhood development, school, work, friends and family utilising the PedsQL measure. They found individuals with DMD often experience lower HRQOL in functional status, fatigue and physical functioning. Abbott and Carpenter (2015) also reported children with a GMD as having more limited social interactions, according to their primary caregiver/parents. This was also consistent with Skyrme (2017) and Uzark et al (2012). Uzark et al. (2012) assessed HRQOL using the PedsQL measure in 117 children living with DMD (6 to 18 years old),

adolescent participants scored significantly lower (13 to 18 years old) than children (between 8 to 12 years) in the physical functioning domain of HRQOL. Uzark et al. (2012) also reported children with DMD scored significantly lower than healthy children for physical and psychosocial domains, including emotional, social, and school functioning by both parent and self-report. This was also consistent with Landfeldt et al. (2015) findings who conducted a cross-population study of HRQOL on participants from Germany, Italy, United Kingdom, and the USA living with a GMDs (770 patient-caregiver pairs), again using the PedsQL measure. While their findings were consistent when compared to the general population, with lower HRQOL observed in physical and social functioning in children with a GMD.

Consistently in previous studies, impairments in HRQOL are noted across several domains of health, including functional status, physical and social domains. The current study (1) identified coping strategies as a significant predictor of total HRQOL scores and (2) the use of social support, problem solving, wishful thinking and self-criticism as coping strategies associated with lower total HRQOL scores. It is important to understand the impact of coping strategies on an individual's health. This study provides sufficient insight into the impact coping strategies have on HRQOL of participants, however, further research is required to better understand why the use of coping strategies is related to lower HRQOL. A negative association between lower total HRQOL and the use of adaptive coping strategies (social support and problem solving) is inconsistent with previous literature. As mentioned earlier, social support was considered an extremely effective coping strategy across several paediatric previous studies, across a range of health-related conditions (Rodgers et al. 2012; Bingen et al., 2012; Smith et al., 2013). Therefore, it was expected to be positively correlated with increased total HRQOL. However, this was not observed in the findings. Previous literature has focused on specific GMDs, such as DMD or BMD; whereas the current study focused on the impact of a range of GMDs, including rare GMDs. The increased use of coping strategies in participants may also be related to more severe symptoms of GMDs, and as a consequence related to lower HRQOL.

This study suggests children used a range of coping strategies. On average, children reported using five different types of coping strategies, with positive strategies more commonly used than negative strategies. A linear association was identified between the increased use of both positive and negative coping strategies and lower total HRQOL scores. Oppenheimer, Krispin, Levy, Ozeri, and Apter (2018) found coping patterns were the

strongest predictor of HRQOL in 273 Israeli children and adolescents (aged 8 to 18 years) diagnosed with a range of health conditions (asthma, diabetes mellitus, or celiac disease). Using the Coping with a Disease Questionnaire (CODI) and the DISABKIDS Chronic Generic Measure (DCGM-37), Oppenheimer and colleagues (2018) found significant inter-correlations among the different strategies, suggesting children used a range of coping strategies. This encouraged the author of the Israeli study to investigate coping patterns instead of individual coping strategies. Oppenheimer and colleagues (2018) reported significantly higher HRQOL in children using effective coping strategies (cognitive restructuring and emotional regulation) compared to children using ineffective coping strategies (such as wishful thinking and avoidance). Another study explored the relationship between types of coping strategies, pain and HRQOL in 104 paediatric Sickle cell disease (SCD) participants (between 8 to 18 years) (Lim et al., 2019). Lim and colleagues (2019) findings suggested emotion-focused coping significantly mediated the association between pain and physical HRQOL. However, no associations were found between psychosocial HRQOL and use of coping strategies. This was inconsistent with the findings of the current study, as significantly lower total HRQOL scores were observed in both positive and negative coping strategy use. A total HRQOL score is the average of the four major subscales, physical functioning, emotional functioning, social functioning, and school functioning. Therefore, higher total scores reflect better HRQOL. The observed differences in the current study's findings may be due to the significant impact GMDs have on an individual's functioning across several domains of health compared to other health conditions. For example, Skyrme (2017) and Morris (1992) suggested children with GMDs are physically limited by pain, discomfort, increased muscle weakness, and a lack of energy; whereas, symptoms for Type 1 diabetes include increased thirst, frequent urination, hunger, and fatigue. Both conditions cannot be cured, however, Type 1 diabetes is often well managed through maintaining blood sugar levels. In contrast, it is extremely difficult to manage symptoms of GMDs and symptom management is often tailored to each specific individual, with the aim of limiting physical pain and discomfort while increasing physical mobility (Lim et al., 2019; Skyrme, 2017).

This study provides a significant contribution to knowledge, it is one of the first studies to consider use of coping strategies in a range of GMDs. Therefore, the findings provide valuable insight into the use and their effectiveness. This has then been discussed and critically analysed referencing previous literature to provide an in depth understanding of the

impact GMDs have on everyday functioning, the strategies that are commonly used, and their perceived effectiveness.

5.4 Strengths and Limitations

The strengths of the current study was its population-based design, which is considered the gold standard of quantitative methods compared to community or hospital-based studies. Therefore, the study is highly quantifiable, and can be repeated by future studies in NZ and globally. Another strength of the current study is its inclusion of children from middle childhood to adolescence, therefore, providing the ability to identify any age-related differences in children's use of coping strategies with living with a GMD. Thirdly, another strength was the inclusivity of all GMDs. This study provides an overview of coping strategy use among all GMDs, as opposed to focusing on the main conditions such as DMD. As a result, study findings offer a broader insight into the coping strategies of children across all GMDs.

A key limitation of this research is the small number of participants involved in the study. However, this was to be expected due to the nature and specific scope of the study. As a result of the specificity of this study, the findings are extremely relevant to children living with GMDs in NZ, and possibly generalisable to other GMD populations across the globe. While the sample population was not large, it included 48 of 159 affected children living in NZ on the point prevalence date. Another possible limitation regarding the current study included missing participant data. Children without Kidcope or PedsQL data available were not included in the current analysis. This may have led to sample bias in the study population, influencing the generalisability if the study findings. Thirdly, a disparity in other ethnic groups other than NZ Europeans was observed in the present study. As a result of this, the findings of this study may largely reflect the coping strategies of NZ European children. Finally, the inclusion of a control group of NZ children would have strengthened the findings of the current study. As it would have allowed us to compare similarities or differences in coping strategies used and their effectiveness in relation to children's HRQOL outcomes.

5.5 Direction for future studies and recommendations for practice

Based on the findings of the current study, several recommendations are offered for future research and practice. First, a notable gap in knowledge was identified in the use of coping strategies in the NZ population, across all ethnicities. Very little recent literature were available. There was also a significant gap in the literature when considering the impact GMDs have on children's HRQOL in NZ. Extensive future research across NZ's key ethnic groups is required to inform clinical practices and to aide those responsible for supporting the lives of children with GMDs in NZ. Further research is required to provide NZ specific symptom management plans for children who are living with a GMD and who are trying to cope with life within the context of changing and potentially worsening impairments in physical, social and/or cognitive domains.

Second, future research could focus on coping strategy use and its influence on specific domains of HRQOL, such as the social, emotional and physical domains. Such research would provide an extension to the findings of the current study by offering a thorough understanding of the domains of HRQOL that are commonly impacted in children living with a GMD. Future research could also include a measure of children's stress, as increased stress may be a confounding factor for the use of coping strategies (Heffer & Willoughby, 2017).

Thirdly, the current study found that social support was perceived as the most effective coping strategy. Positive coping strategies such as cognitive restructuring were commonly used, but perceived as less effective. A reason for coping strategies such as cognitive restructuring not being effective may be due to incorrect use by young individuals. It would be essential to consider integrating the use of positive coping strategies into future treatment plans through the use of social support. An example of integrating use of positive strategies may be through developing group ACT treatment plans for children living with a GMD. This will provide children and their primary caregivers an opportunity to develop social support circles within their community, while still learning adaptive cognitive behavioural management. This will, in turn, coincide with children using adaptive strategies and decrease use of maladaptive coping strategies, such as distraction.

The fourth recommendation is focused on incorporating the findings of this study to develop a future longitudinal cohort study. This could be focused on teaching effective

coping strategies and providing psychoeducation that may be beneficial to children living in NZ with a GMDs. The use of generally effective coping strategies such as social support, emotional regulation, problem-solving and cognitive restructuring may be integrated to enhance children's 'toolbox' for coping with everyday functioning. This can also include a control group of children with no GMDs or another health condition (such as paediatric cancer) to provide comparisons between the long-term effectiveness of coping strategies across various paediatric populations.

5.6 Conclusion

GMDs include a range of genetically acquired disorders that often negatively impact children's HRQOL. Unfortunately, as of yet, there is no absolute cure for GMDs and treatment is often focused on improving quality of life across all domains of health. A gap in the literature was identified regarding the use of coping strategies in children. As a result of this, the current study was conducted to (1) identify the types of coping strategies commonly used by children with GMDs and their effectiveness; (2) how the use of coping strategies may differ depending on the individual demographic characteristics of the child; and (3) highlight any association between coping strategies used and children's self-reported HRQOL. This study also sought to inform future research and the development of new interventions to support children living with a GMD and their families.

Firstly, the findings of this study concluded cognitive restructuring and wishful thinking were the most commonly used coping strategies, whereas, social support and distraction were perceived as the most effective. This was in accordance with previous literature regarding the use of coping strategies and their effectiveness among children and adolescents in a range of paediatric stressors (Bingen et al., 2012; Rodgers et al., 2012; Pereda et al. 2009; Smith et al., 2013). The outcome of this study also informed the conclusion that children commonly used wishful thinking and distraction as a short-term solution to cope with the implications of living with a GMD. In contrast, social support was not commonly used but was perceived as the most effective strategy in both the current study and previous literature by children that used the strategy (Bingen et al., 2012; Smith et al., 2013). Children living with GMDs perceived positive coping strategies to be significantly more effective than negative coping strategies.

In relation to the second aim, there were no significant differences between demographic factors (age, sex and gender) and the use of individual coping strategies. However, adolescent participants used more negative coping strategies compared to children. This is consistent with the progression of GMDs, as children often experience progressively worse physical, cognitive and psychological symptoms with increasing age (Landfeldt et al., 2015). There is conflicting literature regarding the impact of demographic factors and the use of coping strategies, thus the finding of the present study were inconclusive compared to previous findings (Hampel & Petermann, 2005; Donaldson et al., 2000). However, it provided crucial insight into how the use of coping strategies in children with GMDs differ compared to other health conditions.

Finally, HRQOL is significantly impaired in children living with a GMD. However, the current study highlighted conflicting evidence regarding the use of coping strategies and the association it has on self-reported HRQOL. The use of social support, problem solving, wishful thinking and self-criticism coping strategies were associated with lower self-reported HRQOL scores in individuals living with GMDs. The use of both positive and negative coping strategies was also associated with lower HRQOL and accounted for a significant amount of variance explained in self-reported HRQOL. The relationship highlights the differences between the general population and GMD population. The use of positive strategies were not correlated with improved HRQOL. This could be a result of limited access to psychoeducation. Therefore, a recommendation from the current study suggests increased availability of psychoeducation in parallel with physical treatment. This would provide children with a greater depth of understanding to use adaptive coping strategies effectively. If provided in a group setting, this would also positively influence social interactions in both children and their primary caregivers. In conclusion, it is crucial to conduct further research in the use of coping strategies and factors influencing children's HRQOL to increase their quality of life. The findings of the current study contribute to the body of literature in this area and provides new avenues for both the research and development of new interventions for improving children's HRQOL and well-being.

References

Abbott, D., & Carpenter, J. (2015). The things that are inside of you are horrible: Children and young men with Duchenne muscular dystrophy talk about the impact of living with a long-term condition. *Child Care in Practice*, *21*(1), 67–77.

Aflakseir A. (2010). The role of social support and coping strategies on mental health of a group of Iranian disabled war veterans. *Iranian journal of psychiatry*, *5*(3), 102–107.

Amburgey, K., McNamara, N., Bennett, L. R., McCormick, M. E., Acsadi, G., & Dowling, J. J. (2011). Prevalence of congenital myopathies in a representative pediatric united states population. *Annals of Neurology*, *70*(4), 662-665. doi:10.1002/ana.22510

Bann, C. M., Abresch, R. T., Biesecker, B., Conway, K. C., Heatwole, C., Peay, H., … Bolen, J. (2015). Measuring quality of life in muscular dystrophy. *Neurology*, *84*(10), 1034–1042. doi:10.1212/WNL.0000000000001336

Bedel, A., Isik, E., & Hamarta, E. (2014). Psychometric properties of the kidcope in Turkish adolescents. *Eğitim ve Bilim*, *39*(176), 227-235. doi:10.15390/eb.2014.3501

Bingen, K., Kent, M. W., Rodday, A. M., Ratichek, S. J., Kupst, M. J., & Parsons, S. K. (2012). Children's coping with hematopoietic stem cell transplant stressors: Results from the journeys to recovery study. *Children's Health Care*, *41*(2), 145-161. doi:10.1080/02739615.2012.656551

Brace, N., Snelgar, R., & Kemp, R. (2012). *SPSS for psychologists*. London, United Kingdom: Macmillan International Higher Education.

Carroll L. (2013) Problem-focused coping. In: Gellman M.D., Turner J.R. (eds) *Encyclopedia of Behavioral Medicine*. Springer, New York, NY

Carver C. (2013) Coping. in Gellman M.D., Turner J.R. (eds) *Encyclopedia of Behavioral Medicine*. Springer, New York, NY

Carver, C. S. (1997). You want to measure coping but your protocol's too long: consider the Brief COPE. *International Journal of Behavioral Medicine. 4*, 92–100. doi: 10.1207/s15327558ijbm0401_6

Carver, C. S., Scheier, M. F., & Weintraub, J. K. (1989). Assessing coping strategies: A theoretically based approach. *Journal of Personality and Social Psychology*, *56*(2), 267-283. doi:10.1037/0022-3514.56.2.267

Cassandrini, D., Trovato, R., Rubegni, A., Lenzi, S., Fiorillo, C., & Santorelli, F. M. (2017). Congenital myopathies: clinical phenotypes and new diagnostic tools. *Italian Journal of Pediatrics*, *43*(1). doi:10.1186/s13052-017-0419-z

Choi, N. G., Hegel, M. T., Marinucci, M. L., Sirrianni, L., & Bruce, M. L. (2011). Association between participant-identified problems and depression severity in problem-solving therapy for low-income homebound older adults. *International Journal of Geriatric Psychiatry, 27*(5), 491-499. doi:10.1002/gps.2741

Chu-Lien, C. R., (2011). Managing stress and maintaining well-being: Social support, problem-focused coping, and avoidant coping. *J. Couns. Dev. 89*, 338–348. 10.1002/j.1556-6678.2011.tb00098.x

Clarke, D. E., & Jensen, M. A. (1997). The effects of social support, life events, and demographic factors on depression among Māori and Europeans in New Zealand rural, town, and urban environments. *Journal of Community Psychology*, 25, 303-323.

Compas, B. E., Connor-Smith, J. K., Saltzman, H., Thomsen, A. H., & Wadsworth, M. E. (2001). Coping with stress during childhood and adolescence: Problems, progress, and potential in theory and research. *Psychological Bulletin*, *127*(1), 87-127. doi:10.1037//0033-2909.127.1.87

Darin, N., & Tulinius, M. (2000). Neuromuscular disorders in childhood: A descriptive epidemiological study from western Sweden. *Neuromuscular disorders*, *10*(1), 1-9.

de Anda, D., Baroni, S., Boskin, L., Buchwald, L., Morgan, J., Ow, J., Siegel Gold, J., and Weiss, R. (2000). Stress, stressors and coping strategies among high school students. *Child. Youth Serv. Rev. 22,* 441–463.

DePoy, E., & Gitlin, L. N. (2011). *Introduction to research: understanding and applying multiple strategies*. St. Louis, Missouri: Elsevier.

Dijkstra, M. T., & Homan, A. C. (2016). Engaging in rather than disengaging from stress effective coping and perceived control. *Frontiers in psychology, 7*, 1415. doi:10.3389/fpsyg.2016.01415

Donaldson, D., Prinstein, M. J., Danovsky, M., & Spirito, A. (2000). Patterns of children's coping with life stress: Implications for clinicians. *American Journal of Orthopsychiatry, 70*(3), 351-359. doi:10.1037/h0087689

Dudovskiy, J. (2018). Positivism research methodology. Retrieved April 30, 2018, from https://research-methodology.net/research-philosophy/positivism/

Emery, A. E. H. (1987). Duchenne muscular dystrophy. Oxford University Press. Retrieved from http://search.ebscohost.com.ezproxy.aut.ac.nz/

Emery, A. E. (2002). Muscular dystrophy into the new millennium. *Neuromuscular Disorders, 12*(4), 343-349. doi:10.1016/s0960-8966(01)00303-0

Engel, J. M., Kartin, D., & Jaffe, K. M. (2005). Exploring chronic pain in youths with Duchenne Muscular Dystrophy: a model for pediatric neuromuscular disease. *Phys Med Rehabil Clin N Am, 16*(4), 1113-1124.

Folkman, S., & Lazarus, R. S. (1988). *Ways of Coping Questionnaire: Research Edition*. Palo Alto, CA: Consulting Psychologists Press.

Fortes-Ferreira L., Peiró J. M., González-morales G., Martin I. (2006). Work-related stress and well-being: the roles of direct action coping and palliative coping. *Scand. J. Psychol. 47,* 293–302. 10.1111/j.1467-9450.2006.00519.

Gazzard, S. (2004). Muscular dystrophy – the facts. *Neuromuscular Disorders*, *14*(1), 84. doi:10.1016/s0960-8966(03)00113-5

Genetics Home Reference. (2019, April 30). Duchenne and Becker muscular dystrophy. Retrieved May 3, 2019, from https://ghr.nlm.nih.gov/condition/duchenne-and-becker-muscular-dystrophy

Ghriwati, N., Winter, M. A., Everhart, R. S., & Fiese, B. H. (2017). Family functioning and child asthma severity: A bio-behavioral approach. *Families, Systems, & Health*, *35*(4), 439-449.

Halfpenny, P. (2015). *Positivism and sociology: explaining social life*. Oxfordshire, England: Routledge.

Hampel, P., & Petermann, F. (2005). Age and gender effects on coping in children and adolescents. *Journal of Youth and Adolescence*, *34*(2), 73-83. doi:10.1007/s10964-005-3207-9

Heffer, T., & Willoughby, T. (2017). A count of coping strategies: A longitudinal study investigating an alternative method to understanding coping and adjustment. *PloS one*, *12*(10). doi:10.1371/journal.pone.0186057

Hope, D. A., Burns, J. A., Hayes, S. A., Herbert, J. D., & Warner, M. D. (2007). Automatic thoughts and cognitive restructuring in cognitive behavioral group therapy for social anxiety disorder. *Cognitive Therapy and Research*, *34*(1), 1-12. doi:10.1007/s10608-007-9147-9

Hughes, M., Hicks, E., Nevin, N., & Patterson, V. (1996). The prevalence of inherited neuromuscular disease in Northern Ireland. *Neuromuscular Disorders*, *6*(1), 69-73. doi:10.1016/0960-8966(94)00017-4

Huml, R. A. (2015). Muscular dystrophy: A concise guide. Basingstoke, England: Springer.

Hunt, A., Carter, B., Abbott, J., Parker, A., Spinty, S., & deGoede, C. (2016). Pain experience, expression and coping in boys and young men with Duchenne Muscular Dystrophy - A pilot study using mixed methods. *European Journal of Paediatric Neurology, 20*(4), 630–638.

Jones, K. M., O'Grady, G., Rodrigues, M. J., Ranta, A., Roxburgh, R. H., & Love, D. R. (2018). Impacts for children living with genetic muscle disorders and their parents – Findings from a population-based study. *Journal of Neuromuscular Diseases, 5*(3), 341-352. doi:10.3233/jnd-170287

Jose, P. E., & Schurer, K. (2009). Cultural differences in coping among New Zealand adolescents. *Journal of Cross-Cultural Psychology, 41*(1), 3-18. doi:10.1177/0022022109348783

Kim, J., Jeong Yeob Han, Shaw, B., McTavish, F., & Gustafson, D. (2010). The roles of social support and coping strategies in predicting breast cancer patients' emotional well-being. *Journal of Health Psychology, 15*(4), 543-552. doi:10.1177/1359105309355338

Kohler, M., Clarenbach, C. F., Bahler, C., Brack, T., Russi, E. W., & Bloch, K. E. (2009). Disability and survival in Duchenne muscular dystrophy. *Journal of Neurology, Neurosurgery & Psychiatry, 80*(3), 320-325. doi:10.1136/jnnp.2007.141721

Krohnea, H. W. (2002). Stress and coping theories. Retrieved from http://userpage.fu-berlin.de/schuez/folien/Krohne_Stress.pdf

Kuo, D., Bird, T., & Tilford, J. (2011). Associations of family-centered care with health care outcomes for children with special health care needs. *Maternal and child health journal, 15*(6), 794-805.

Landfeldt, E., Lindgren, P., Bell, C. F., Guglieri, M., Straub, V., Lochmüller, H., & Bushby, K. (2015). Health-related quality of life in patients with Duchenne muscular dystrophy: a multinational, cross-sectional study. *Developmental Medicine & Child Neurology, 58*(5), 508-515. doi:10.1111/dmcn.12938

Law, M., Hanna, S., Anaby, D., Kertoy, M., King, G., & Xu, L. (2014). Health-related quality of life of children with physical disabilities: a longitudinal study. *BMC Pediatrics*, *14*(1). doi:10.1186/1471-2431-14-26

Leech, N. L., Barrett, K. C., & Morgan, G. A. (2014). *IBM SPSS for intermediate statistics: Use and interpretation* (5th ed.). London, England: Routledge.

Lim, C. S., Karlson, C., Edmond, S. N., Welkom, J. S., Osunkwo, I., & Cohen, L. L. (2019). Emotion-focused avoidance coping mediates the association between pain and health-related quality of life in children with sickle cell disease. *Journal of Pediatric Hematology/Oncology*, *41*(3), 194-201. doi:10.1097/mph.0000000000001429

Lovering, R. M., Porter, N. C., & Bloch, R. J. (2005). The muscular dystrophies: From genes to therapies. *Physical Therapy*, *85*(12), 1372–1388. doi:10.1093/ptj/85.12.1372

Lue, Y., Chen, S., & Lu, Y. (2016). Quality of life of patients with Duchenne muscular dystrophy from adolescence to young men. *Disability and Rehabilitation*, *39*(14), 1408-1413. doi:10.1080/09638288.2016.1196398

Moos, R. H., & Schaefer, J. A. (1993). Coping resources and processes current concepts and measures. In *Handbook of stress: Theoretical and clinical aspects* (2nd ed.). New York: Free Press.

Muscular Dystrophy Association. (2019, February). Duchenne muscular Dystrophy (DMD). Retrieved May 17, 2019, from https://www.mda.org

National Institute of Neurological Disorders and Stroke (NINDS). (2019, March 27). Congenital myopathy. Retrieved October 13, 2019, from https://www.ninds.nih.gov/disorders/all-disorders/congenital-myopathy

National Organization for Rare Disorders (NORD). (2015, February 11). Central core disease. Retrieved October 13, 2019, from https://rarediseases.org/rare-diseases/central-core-disease/

Norwood, F. L., Harling, C., Chinnery, P. F., Eagle, M., Bushby, K., & Straub, V. (2009). Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. *Brain, 132*(11), 3175-3186. doi:10.1093/brain/awp236

Oppenheimer, S., Krispin, O., Levy, S., Ozeri, M., & Apter, A. (2018). The impact of coping patterns and chronic health conditions on health-related quality of life among children and adolescents. *European Journal of Pediatrics, 177*(6), 935-943. doi:10.1007/s00431-018-3146-6

Pallant, J. (2016). *SPSS Survival Manual: A step by step guide to data analysis using IBM SPSS*. Crows Nest, Australia: Allen & Unwin.

Parker, J. D. A., and Endler, N. S. (1992). Coping with coping assessment: A critical review. *Eur. J. Pers. 6*, 321–344. doi: 10.1002/per.2410060502

Pereda, N., Forns, M., Kirchner, T., & Muñoz, D. (2009). Use of the Kidcope to identify socio-economically diverse Spanish school-age children's stressors and coping strategies. *Child: Care, Health and Development, 35*(6), 841-850. doi:10.1111/j.1365-2214.2009.00991.x

Power, T. G. (2004). Stress and coping in childhood: The parents' role. *Parenting, 4*(4), 271-317. doi:10.1207/s15327922par0404_1

Pretzlik, U., & Sylva, K. (1999). Paediatric patients' distress and coping: an observational measure. *Archives of Disease in Childhood, 81*(6), 528-530. doi:10.1136/adc.81.6.528

Read, J., Kinali, M., Muntoni, F., Weaver, T., & Garralda, M. E. (2011). Siblings of young people with Duchenne muscular dystrophy: A qualitative study of impact and coping. *European Journal of Paediatric Neurology, 15*(1), 21-28. doi:10.1016/j.ejpn.2010.07.006

Reinoso, M., Pereda, N., Van den Dries, L., & Forero, C. G. (2013). Internationally adopted children's general and adoption-specific stressors, coping strategies and psychological adjustment. *Child & Family Social Work*, *21*(1), 1-13. doi:10.1111/cfs.12099

Rodgers, C., Norville, R., Taylor, O., Poon, C., Hesselgrave, J., Gregurich, M. A., & Hockenberry, M. (2012). Children's coping strategies for chemotherapy-induced nausea and vomiting. *Oncology Nursing Forum*, *39*(2), 202–209. Retrieved from http://search.ebscohost.com.ezproxy.aut.ac.nz/login.aspx?direct=true&db=ccm&AN=104525960&site=eds-live

Rodrigues, M. J., O'Grady, G. L., Hammond-Tooke, G., Kidd, A., Love, D. O., Baker, R. K., & Roxburgh, R. H. (2017). The New Zealand neuromuscular disease patient registry; five years and a thousand patients. *Journal of neuromuscular diseases*, *4*(3), 183–188. doi:10.3233/JND-170240

Roecker, C. E., Dubow, E. F., & Donaldson, D. (1996). Cross-situational patterns in children's coping with observed interpersonal conflict. *Journal of Clinical Child Psychology*, *25*(3), 288-299. doi:10.1207/s15374424jccp2503_5

Rubin, M. (2019, January). Congenital Myopathies. Retrieved October 13, 2019, from https://www.msdmanuals.com/en-nz

Sarantakos, S. (2013). *Social research*. Basingstoke: Palgrave Macmillan.

Smith, T. F., Russell, H. F., Kelly, E. H., Mulcahey, M. J., Betz, R. R., & Vogel, L. C. (2013). Examination and measurement of coping among adolescents with spinal cord injury. *Spinal Cord*, *51*(9), 710-714. doi:10.1038/sc.2013.65

Skinner, E. A., Edge, K., Altman, J., & Sherwood, H. (2003). Searching for the structure of coping: A review and critique of category systems for classifying ways of coping. *Psychological Bulletin*, *129*(2), 216-269. doi:10.1037/0033-2909.129.2.216

Skinner, E. A., & Zimmer-Gembeck, M. J. (2006). The development of coping. *Annual Review of Psychology*, *58*, 119-144. doi:10.1007/978-3-319-41740-0

Skyrme, S. (2017). In and on their own terms: children and young people's accounts of life with Duchenne muscular dystrophy. *Child Care in Practice, 23*(1), 77–89. https://doi-org.ezproxy.aut.ac.nz/10.1080/13575279.2016.1158152

Spirito, A., Stark, L. J., & Williams, C. (1988). Development of a brief coping checklist for use with pediatric populations. *Journal of Pediatric Psychology, 13*(4), 555-574. doi:10.1093/jpepsy/13.4.555

Stanisławski, K. (2019). The coping circumplex model: An integrative model of the structure of coping with stress. *Frontiers in Psychology, 10*. doi:10.3389/fpsyg.2019.00694

Strehle, E. (2009). Long-term management of children with neuromuscular disorders. *Jornal de Pediatria, 85*(5), 379-384. doi:10.2223/jped.1929

Sveinbjornsdottir, S., & Thorsteinsson, E. B. (2014). Psychometric properties of the measure of adolescent coping strategies (MACS). *Psychology, 05*(02), 142-147. doi:10.4236/psych.2014.52022

Theadom, A., Rodrigues, M., Poke, G., O'Grady, G., Love, D., Parmar, P., … Hammond-Tooke, G. (2019). A nationwide, population-based prevalence study of genetic muscle disorders. *Neuroepidemiology, 52*(4), 128-135. doi:10.1159/000494115

Todd, J. J., Razaqyar, M. S., Witherspoon, J. W., Lawal, T. A., Mankodi, A., Chrismer, I. C., … Meilleur, K. G. (2018). Novel variants in individuals with RYR1-related congenital myopathies: genetic, laboratory, and clinical findings. *Frontiers in Neurology, 9*. doi:10.3389/fneur.2018.00118

Uzark, K., King, E., Cripe, L., Spicer, R., Sage, J., Kinnett, K., … Varni, J. W. (2012). Health-related quality of life in children and adolescents with duchenne muscular dystrophy. *Pediatrics, 130*(6), 1559-1566. doi:10.1542/peds.2012-0858

Varni, J., Seid, M., & Rode, C. (1999). The PedsQL: measurement model for the pediatric quality of life inventory. *Medical Care, 37*(2), 126-139.

Varni, J. W., Seid, M., & Kurtin, P. S. (2001). PedsQL 4.0: Reliability and validity of the pediatric quality of life inventory version 4.0 generic core scales in healthy and patient populations. *Medical Care, 39*(8), 800-812. doi:10.1097/00005650-200108000-00006

Varni, J. W., Waldron, S. A., Gragg, R. A., Rapoff, M. A., Bernstein, B. H., Lindsley, C. B., & Newcomb, M. D. (1996). Development of the waldron/varni pediatric pain coping inventory. *Pain, 67*(1), 141-150. doi:10.1016/0304-3959(96)03077-1

Vetter, T. R., McGwin, G., Bridgewater, C. L., Madan-Swain, A., & Ascherman, L. I. (2013). Validation and clinical application of a biopsychosocial model of pain intensity and functional disability in patients with a pediatric chronic pain condition referred to a subspecialty clinic. *Pain Research and Treatment, 2*(1), 1-12. doi:10.1155/2013/143292

Viecili, M. A., & Weiss, J. A. (2015). Reliability and validity of the pediatric quality of life inventory with individuals with intellectual and developmental disabilities. *American Journal on Intellectual and Developmental Disabilities, 120*(4), 289-301. doi:10.1352/1944-7558-120.4.289

Webb, C. L. (2005). Parents' perspectives on coping with duchenne muscular dystrophy. *Child: Care, Health and Development, 31*(4), 385-396. doi:10.1111/j.1365-2214.2005.00518.x

Wei, Y., Speechley, K., & Campbell, C. (2015). Health-related quality of life in children with duchenne muscular dystrophy. *Journal of Neuromuscular Diseases, 2*(3), 313-324. doi:10.3233/jnd-150071

Wertlieb, D., Weigel, C., & Feldstein, M. (1987). Stress, social support, and behavior Symptoms in middle childhood. *Journal of Clinical Child Psychology, 16*(3), 204-211. doi:10.1207/s15374424jccp1603_4

Zalk, R., Clarke, O. B., Des Georges, A., Grassucci, R. A., Reiken, S., Mancia, F., … Marks, A. R. (2014). Structure of a mammalian ryanodine receptor. *Nature*, *517*(7532), 44-49. doi:10.1038/nature13950

Zatz, M., Passos-Bueno, M. R., & Vainzof, M. (2016). Neuromuscular disorders: genes, genetic counseling and therapeutic trials. *Genetics and Molecular Biology*, *39*(3), 339-348. doi:10.1590/1678-4685-gmb-2016-0019

Appendices

Appendix A: AUTEC ethics approval

AUTEC
SECRETARIAT

11 September 2014

Alice Theadom
Faculty of Health and Environmental Sciences

Dear Alice

Ethics Application: **14/296 Prevalence and impact of genetic muscle disorders in New Zealand (MD-Prev).**

Thank you for submitting your application for ethical review to the Auckland University of Technology Ethics Committee (AUTEC. I am pleased to confirm that the Chair and I have approved your ethics application for three years until 11 September 2017

As part of the ethics approval process, you are required to submit the following to AUTEC:

- A brief annual progress report using form EA2, which is available online through http://www.aut.ac.nz/researchethics. When necessary this form may also be used to request an extension of the approval at least one month prior to its expiry on 11 September 2017;
- A brief report on the status of the project using form EA3, which is available online through http://www.aut.ac.nz/researchethics. This report is to be submitted either when the approval expires on 11 September 2017 or on completion of the project;

It is a condition of approval that AUTEC is notified of any adverse events or if the research does not commence. AUTEC approval needs to be sought for any alteration to the research, including any alteration of or addition to any documents that are provided to participants. You are responsible for ensuring that research undertaken under this approval occurs within the parameters outlined in the approved application.

AUTEC grants ethical approval only. If you require management approval from an institution or organisation for your research, then you will need to obtain this.

To enable us to provide you with efficient service, we ask that you use the application number and study title in all correspondence with us. If you have any enquiries about this application, or anything else, please do contact us at ethics@aut.ac.nz.

All the very best with your research,

Kate O'Connor
Executive Secretary
Auckland University of Technology Ethics Committee

Auckland University of Technology Ethics Committee
WA505D Level 5 WA Building City Campus
Private Bag 92006 Auckland 1142 Ph: +64-9-921-9999 ext 8316 email ethics@aut.ac.nz

Appendix B: Child and adolescent information sheet and consent form

1

Child and Adolescent Information Sheet

Study title:	**Prevalence and impact of genetic muscle disorders in New Zealand**		
Locality:	**New Zealand**	Ethics committee ref.:	**14/NTB/118**
Lead investigator:	**Alice Theadom**	Contact phone number:	**0800 MDPREV**

WHAT IS THE STUDY ABOUT?

You are invited to take part in a study that is looking at how many people have a genetic muscle disorder in New Zealand.

We would like to support people who have a genetic muscle disorder by finding out how people are doing and what help they are currently receiving.

Whether or not you take part is your choice. If you do want to take part now, but change your mind later, you can tell us that you want to stop at any time and that will be ok.

You do not have to decide today whether or not you want to take part. Before you decide you can talk to your family, whānau or friends.

WHO IS INVOLVED?

We are a team of people who work in universities, health care services, and community organisations in New Zealand.

WHAT WILL I NEED TO DO?

If you would like to take part, we will arrange a time to come and talk to you. When a researcher comes to visit you, you will be able to ask any other questions you may have about the study.

They will then ask you some questions about how you are feeling and how you find doing activities at home and outside.

We would also like to ask your parent, or a person who looks after you, some questions about how you and they have been doing.

WHAT ELSE SHOULD I KNOW?

If you feel uncomfortable or start to feel upset, please tell the person speaking with you. You can take a break if you would like to or we can ask you some different questions.

You do not have to answer anything you do not want to. If you would like us to stop talking to you or person who looks after you at any point, that's okay, please just tell us you want us to stop talking to them.

To say thank you for taking part, we will give you a $20 Westfield voucher once you have finished answering all the questions.

We will keep everything you tell us private. Your name will not be put on the paper with your answers on so no one will know what you have said. But if we think that you, or someone else, might not be

safe, we will have to tell someone who can help us to keep you safe. We will tell you first if we feel we need to do this.

WHAT HAPPENS AFTER WARDS?

When we look at what everyone has told us, we will write about what we have found.

We will also talk to doctors and people who run health care services to help them to help people with genetic muscle disorders better.

After the study has finished we will keep all your information locked in a cupboard at the Auckland University of Technology in Auckland. We will keep your information for 10 years after you have turned 16 years of age before it will be destroyed.

WHO DO I CONTACT FOR MORE INFORMATION OR IF I HAVE CONCERNS?

If you have any worries or questions please contact us.

Kerry Walker, Study Manager
Telephone: 0800 MDPREV (637738)
Mobile: 0212458597
Email: kwalker@aut.ac.nz

If you want to talk to someone who isn't involved with the study, you can contact the advocacy team on:

Phone: 0800 555 050
Fax: 0800 2 SUPPORT (0800 2787 7678)
Email: advocacy@hdc.org.nz

You can also contact the people that approved this study on:

Phone: 0800 4 ETHICS
Email: hdecs@moh.govt.nz

Thank you for reading about this study
You can keep this information.

4

Registration Number:	Participant Initials:	Date of Birth:

Child/Adolescent Consent Form

Please tick if you agree with the following

I have read and I understand the Information Sheet.

I have had the opportunity to talk to others to help me ask questions and understand the study.

I know who to contact if I have any questions about the study in general.

Declaration by participant:
I agree to take part in this study.

Participant's name: _____

Signature: _____ Date: _____

Declaration by member of research team:

I have explained the study to the participant and answered the participant's questions about it. I believe that the participant understands the study and has given their assent to participate.

Researcher's name: _____

Signature: _____ Date: _____

Appendix C: Parent/legal guardian information sheet and consent form

1

Parent/Legal Guardian Information Sheet

Study title:	**Prevalence and impact of genetic muscle disorders in New Zealand**		
Locality:	**New Zealand**	Ethics committee ref.:	**14/NTB/118**
Lead investigator:	**Alice Theadom**	Contact phone number:	**0800 MDPREV**

You are invited to take part in a study that is looking at how many people are affected by genetic muscle disorders in New Zealand. We would also like to find out how people and their significant others are affected by these conditions.

Whether or not you take part is your choice. If you don't want to take part, you don't have to give a reason, and it won't affect the care you or your child receives. If you do want to take part now, but change your mind later, you can pull out of the study at any time.

This information sheet will help you decide if you'd like to take part. It sets out why we are doing the study, what your participation would involve, what the benefits and risks to you might be, and what would happen after the study ends. We will go through this information with you and answer any questions you may have. You do not have to decide today whether or not you will participate in this study. Before you decide you may want to talk about the study with other people, such as family, whānau, friends, or healthcare providers. Feel free to do this.

If you agree to take part in this study, you will be asked to sign the consent form on the last page of this document. You will be given a copy of both the Participant Information Sheet and the Consent Form to keep.

This document is 7 pages long, including the Consent Form. Please make sure you have read and understood all the pages.

WHAT IS THE PURPOSE OF THE STUDY?

We are a team of researchers who work in universities, hospitals and community organisations across New Zealand, with an interest in supporting people with neuromuscular conditions.
The purpose of this study is to find out how many people are affected by genetic muscle disorders in New Zealand. We would also like to find out how peoples' everyday lives are affected and to explore the impact on those around them. We hope that by finding out this information and identifying areas of unmet need, will help us to improve the support and treatment children, adults and their family/whānau receive. Even if your child is not experiencing any effects from their condition, this is just as important for us to know as if they do.

This study is funded by the Health Research Council of New Zealand.

If you have any questions about the study please contact the Study Manager, Kerry Walker:

Telephone: 0800 MDPREV (637738)
Mobile: 021 2458597
E mail: kwalker@aut.ac.nz

This study has been approved by the Health and Disability Ethics Committee reference: 14/NTB/118

WHAT WILL MY PARTICIPATION IN THE STUDY INVOLVE?

Your child has been identified as someone who may have a genetic muscle disorder. We are inviting everyone with a genetic muscle disorder in New Zealand to participate in this study (about 1200 people). This will enable us to explore the services and supports people receive across the country.

If you would like to take part in this study, we would like to arrange a time to come and talk to you and your child. You will be asked questions about how they find completing everyday activities, socializing and about any symptoms they experience. We would then like to ask some questions about your own quality of life. All researchers who will be asking these questions have been specially trained for this project. We can arrange for a member of the team to visit you and your child at your home or other convenient location such as at your local GP surgery, at the child's school or a private room in a public library. You can also complete some questions yourself on a questionnaire or over the phone if you prefer.

When a researcher comes to visit you, you will have the opportunity to ask any questions you may have about the study. If you are happy to take part, you will be invited to sign the consent form. The assessment should take about one hour to complete.

You will be able to have support people with you during the assessment and we can complete the assessment over several sessions if you prefer.

We aim to finish collecting data for this study by the end of March 2016.

WHAT ARE THE POSSIBLE BENEFITS AND RISKS OF THIS STUDY?

Taking part in this study will take some of your time and require you to answer a series of questions. There are no known risks caused by this study, however you may feel uncomfortable or embarrassed by some of the questions. You do not have to answer any questions you do not wish to do so. All our researchers have received training in administering these assessments and working with people who have a genetic muscle disorder.

Your (or your child's) usual medical care will not be affected in any way by participating in the study, or withdrawing from the study at any stage. Your (and your child's) participation in this study will be stopped should any harmful effects appear or if the doctor feels it is not in your best interests to continue. Similarly your doctor may at any time provide you (or your child) with any other treatment he/she considers necessary.

As part of the study we will be working with health care and service providers about the findings of the study. We aim to identify areas of unmet need for people living with these conditions to improve service delivery. We will also be in contact with your GP or neurologist about your child's diagnosis and if any information that may be of benefit to you and your child emerges during the study we will let them (and you) know.

WHO PAYS FOR THE STUDY?

There should be no direct costs to you in taking part in this study.

A $20 food/fuel voucher will be provided to you after completion of the assessment (your child will also be given a $20 voucher on completion of their assessment) in acknowledgment of the contributions you and your child have made to this study.

Assessments will be completed at your home or other accessible location. If for some reason you need to travel for the assessment, your mileage or costs will be reimbursed.

WHAT IF SOMETHING GOES WRONG?

It is unlikely that you will be at risk of harm from taking part in this study. If something goes wrong, please contact the study manager as soon as possible on 0800 MDPREV (637738).

WHAT ARE MY RIGHTS?

Your participation is entirely voluntary and you will be able to withdraw from the study at any time without experiencing any disadvantage.

The study files and all other information that you provide will remain strictly confidential, unless information is revealed that indicates you, your child or someone else is at risk. The answers to your questions will be stored separately to any document that has your name and contact details on.

No material that could personally identify you will be used in any reports or discussions about this study. As we will be collecting a lot of valuable information as part of the study we would like to share anonymised data will other international researchers following completion of the study. However we will only share your and your child's data if you agree for us to do so, otherwise your data will be removed before it is shared outside of the study team.

You will be able to access your information collected as part of the study if you wish to do so. If any information that may be of benefit to you emerges during the study we will contact you to let you know

WHAT HAPPENS AFTER THE STUDY OR IF I CHANGE MY MIND?

Upon completion of the study your and your child's information will be stored for 16 years in a locked cabinet at AUT University in Auckland by the lead investigator (Dr Alice Theadom). All computer records will be password protected. Any identifying information will not be shared outside of the research team without seeking your permission.

After 16 years all your electronic information will be deleted and paper forms will be shredded and destroyed with the university confidential waste. Your child's information will be stored for 10 years after they have turned 16 years of age.

After we have looked at all the data we will send you a summary of results if you would like to receive them.

WHO DO I CONTACT FOR MORE INFORMATION OR IF I HAVE CONCERNS?

If you have any questions, concerns or complaints about the study at any stage, you can contact:

Kerry Walker, Study Manager
Telephone number: 0800 MDPREV (637738)
Email: kwalker@aut.ac.nz

If you want to talk to someone who isn't involved with the study, you can contact an independent health and disability advocate on:

Phone: 0800 555 050
Fax: 0800 2 SUPPORT (0800 2787 7678)
Email: advocacy@hdc.org.nz

For Maori health support please contact :

Te Puna Oranga (Waikato DHB Maori Health Unit),
Hockin Building, Level 1, Pembroke St, P.O.Box 934, Hamilton.
Ph: (07) 834 3644. Fax: (07) 834 3619.

You can also contact the health and disability ethics committee (HDEC) that approved this study on:

Phone: 0800 4 ETHICS
Email: hdecs@moh.govt.nz

Please keep this for your information.
Thank you for interest in this study

6

Registration Number:	Participant Initials:	Date of Birth:

Parent/Guardian Consent Form

If you need an INTERPRETER, please tell us.

I have read, or have had read to me in my first language, and I understand the Parent/Legal Guardian Information Sheet.

I have been given sufficient time to consider whether or not to participate in this study.

I have had the opportunity to use a legal representative, whanau/ family support or a friend to help me ask questions and understand the study.

I am satisfied with the answers I have been given about the study and I have a copy of this consent form and information sheet.

I understand that taking part in this study is voluntary (my choice) and that I may withdraw from the study at any time without this affecting my medical care.

I consent to the research staff collecting and processing my information, including information about my health from medical records and health databases.

I consent to the research staff contacting my child's teacher for information about their progress at school

If I decide to withdraw from the study, I agree that the information collected about me (and my child) up to the point when I withdraw may continue to be processed.

I consent to my own or my child's GP or treating clinician being informed about my participation in the study and of any significant abnormal results obtained during the study.

MUSCULAR DYSTROPHY Association of New Zealand Inc. Te Rōpū Mate Horokiwa o Aotearoa

AUT UNIVERSITY

7

I agree to an approved auditor appointed by the New Zealand Health and Disability Ethic Committees, or any relevant regulatory authority or their approved representative reviewing my relevant medical records for the sole purpose of checking the accuracy of the information recorded for the study.

I understand that my (and my child's) participation in this study is confidential and that no material, which could identify me (or my child) personally, will be used in any reports on this study.

I know who to contact if I have any questions about the study in general.

I understand my responsibilities as a study participant.

I wish to receive a summary of the results from the study.	Yes ☐	No ☐
I agree to my own and my child's anonymised data being shared with researchers overseas for further research into neuromuscular conditions	Yes ☐	No ☐

Declaration by Parent/Legal Guardian:
I hereby consent for myself and my child, (NAME) _____to take part in this study.

Parent/Legal Guardian's name: _____

Signature: _____ Date: _____

Declaration by member of research team:

I have given a verbal explanation of the research project to the participant, and have answered the participant's questions about it.

I believe that the parent/legal guardian understands the study and has given informed consent to participate.

Researcher's name: _____

Signature: _____ Date: _____