THE EXPERIENCES OF EMERGING ADULTS WITH JUVENILE ONSET MUSCULAR DYSTROPHY: IMPLICATIONS FOR COUNSELORS

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By

Kelsey A. Jager

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A dissertation written by

Kelsey A. Jager

B.A., Kent State University, 2006

M.Ed., Kent State University, 2010

Ph.D., Kent State University, 2019

Approved by

_____, Co-director, Doctoral Dissertation Committee
Jason McGlothlin

_____, Co-director, Doctoral Dissertation Committee
Martin Jencius

_____, Member, Doctoral Dissertation Committee
Phillip Rumrill

Accepted by

_____, Director, School of Lifespan Development and Educational Sciences
Mary Dellmann-Jenkins

_____, Dean, College and Graduate School of Education, Health, and Human Services
James Hannon

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THE EXPERIENCES OF EMERGING ADULTS WITH JUVENILE ONSET MUSCULAR DYSTROPHY: IMPLICATIONS FOR COUNSELORS (255 pp.)

Co-directors of dissertation: Jason McGlothlin, Ph.D.
Marty Jencius, Ph.D.

Emerging adults living with juvenile onset muscular dystrophy (JOMD) are faced with challenging developmental tasks while adjusting to slow, untimely, progressive physical declines. This qualitative study examined the perspectives of eight emerging adults between 23–29 years old with limb-girdle MD (LGMD) or Emery-Dreifuss MD (EDMD)—collectively termed juvenile onset MD (JOMD)—for the purpose of increasing understanding of the experience the subjects afflicted with the disease undergo. Compared to those diagnosed with MD in early childhood, those with juvenile onset have a slower progression which becomes more disabling during adolescence and emerging adulthood. Utilizing a phenomenological framework, five themes emerged from this study. Five themes captured the essential and common elements in the cases of the eight participants in the study and are as follows: (a) JOMD expands into consciousness with continued loss; (b) the stress of experiencing increased visibility; (c) the struggle of justifying symptoms of MD to both oneself and others; (d) evolution of disease, evolution of negative emotion; and (e) fear of the future. Participants describe living with JOMD to be like "hitting a brick wall" time and time again, as physical symptoms become increasingly more restrictive. The mental and emotional effort required to live with MD during adolescence and emerging adulthood is taxing and limits

work and social opportunities. Implications for counseling are discussed, in addition to limitations of the study and recommendations for future research.

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CHAPTER I

INTRODUCTION AND REVIEW OF THE LITERATURE

Imagine, for a moment, what it would be like to be diagnosed with a chronic, incurable, progressive muscle disease that will slowly erode your strength and ability to move your body. Within 10 years, walking will be challenging, and you will fall easily. If no one is around to help you get up, it is possible that you will be stuck, unable to get back onto your feet until you call for help or crawl to assistance. For some, it is possible to have heart problems or even a sudden heart attack. You are informed that strenuous exercise will accelerate the muscle degeneration, but so will a sedentary lifestyle (Emery, 2002). Imagine confronting this reality at the age of 12. This is the challenge at hand for those living with juvenile onset muscular dystrophy (JOMD).

Muscular dystrophy (MD) is an umbrella term for hereditary, chronic, incurable diseases that weaken skeletal muscle and can compromise cardiac and respiratory functioning (Emery, 2002). Since the time the disease was discovered in 1830, advancements in medicine and technology have helped medical professionals gain important knowledge that has led to a sophisticated understanding of various forms of the disease, subsequently minimizing the time between symptom onset and diagnosis and classification of different conditions by distinct genetic defects (Gawlik, 2018). Compared to those who have muscle weakening as a secondary disturbance of other tissues (e.g., nervous system in multiple sclerosis, amyotrophic lateral sclerosis), the muscular dystrophies stem from direct abnormalities of the muscle structure and function (Gawlik, 2018).

Persons with MD represent a small group in a growing population of individuals with chronic disease. In recent decades, the prevalence of chronic disease has increased dramatically. According to the Centers for Disease Control and Prevention (CDC), one out of every two adults has one chronic condition, and four in 10 adults have two or more chronic diseases (2018). The cost of chronic disease is high. It is the leading cause of death and disability among Americans and carries an annual health care cost of $3.3 trillion (CDC, 2018). The most common chronic conditions include heart disease, stroke, chronic lung disease, cancer, Alzheimer's disease, chronic kidney disease, and diabetes (CDC, 2018).

As in the adult population, the prevalence of chronic disease during childhood and adolescence has increased steadily over the past decade. Youth with chronic diseases are at risk for a range of psychological and social adjustment problems that interfere with optimal development (Hauser-Cram, Krauss & Kersh, 2009). Adolescents and emerging adults living with JOMD must deal with ongoing and progressive muscle weakness that impacts day-to-day life, while facing the same developmental tasks and hardships as healthy peers. Given the myriad of risk factors for adjustment problems, it is important to first assess and describe the experience from first-hand accounts. Yet, very little literature exists examining the perspectives of young adults with MD. This research study seeks to understand and describe the experiences of emerging adults living with a form of JOMD.

I came to this study with the following questions:

1. How do emerging adults perceive and describe their experiences of living with JOMD, beginning with first memories of the disease up until now?
2. What counseling interventions could promote improved adjustment to living with JOMD and contribute to both short and long-term wellbeing?

I asked the questions in this way, because my broader goal is to understand the complexity of the psychological and social context of emerging adults who are growing up with a form of JOMD. Of special interest is how progression of the disease is experienced, and the impact it has during adolescence and emerging adulthood.

The profession of counseling is dedicated to establishing "a professional relationship that empowers diverse individuals, families, and groups to accomplish mental health, wellness, education, and career goals" (American Counseling Association [ACA] Code of Ethics, 2014). Counselors are committed to promoting mental health and human functioning by assisting others with stressful transitions across the lifespan. With an emphasis on theory, science, psychology, and a person-centered approach, counselors are well qualified to contribute to the care of those living with chronic illness (Brady-Amoon & Keefe-Cooperman, 2017). As chronic disease is becoming the nation's leading healthcare concern, accounting for 83% of healthcare spending (Anderson, 2004) and 7 out of every 10 fatalities (CDC, 2018), counseling needs to find its place among the new gold standard of treatment—interdisciplinary care (Anderson, 2004).

Counselors could be integral in assisting emerging adults with juvenile onset MD understand the disease, process the reality of vanishing physical strength while helping them integrate the disease into identity development and future planning. While medical

doctors focus on the physical symptoms of the disease, counselors are better prepared to address the mental and emotional consequences. Furthermore, counselors have specialized training in prevention and early identification of mental health problems and issues with life transitions (Brady-Amoon & Keefe-Cooperman, 2017), all useful components of a multidisciplinary treatment team approach. Mental health counselors are trained to offer therapeutic interventions that improve the health, happiness, and resilience of others. However, before effective services can be delivered, an investigation into the lives, challenges, and needs of those with JOMD must be completed.

The research is also prompted by the dearth of literature on young adults living with slowly progressive muscular diseases. The current psychosocial literature on MD is mostly composed of heterogeneous samples of adults (ages 18–70) diagnosed with different forms of the disease with wide ranging physical presentations. Developmental considerations are virtually nonexistent in this literature. Consequently, professionals in the field of medical and mental health are left with treatment approaches that have been created outside the lifespan and illness contexts of the individuals to which they are to be applied.

The developmental theory of emerging adulthood provides further justification for this study. The transition into adulthood is a time of intense instability and focused personal development for all young people, but the process is more challenging for those with chronic childhood disease (Arnett, 2000; Waldboth, Patch, Mahrer-Imhof, & Metcalfe, 2016). Individuals in their emerging adulthood are encountering the most unstable period of their lifespan, marked by identity exploration, rapid changes in

relationships, jobs and housing, high degree of internal focus, feelings of being neither an adolescent or an adult, and high expectations for themselves in the future (Arnett, 2000). Those who are simultaneously dealing with a chronic disease show higher rates of psychopathology (Bernstein, Stockwell, Gallagher, Rosenthal, & Soren, 2013; Kiviruusu, Huurre, & Aro, 2007; Packham, 2004) and lower likelihood of successful transition to adulthood (Pinquart, 2014). Accessing healthcare becomes more difficult due to aging out of longstanding pediatric physicians' services and eventual termination of parental insurance coverage (Colver et al., 2013). Furthermore, mental health counselors have little empirical literature to incorporate into understanding the psychological experience of their clients diagnosed with MD.

My quest for more informed treatment approaches is as much prompted by the impact the illness has had on my family as it is by my passion for the counseling profession. Both my brother and I live with Limb Girdle MD (LGMD2a) and it has taken a toll on us, my parents, and siblings. In our case, little attention was, per doctor recommendation, given to full diagnosis or treatment planning, because it was thought that nothing could be done to slow or stop the progression. We were left to discover for ourselves what it really meant to live with MD. While I am grateful for the support that we were eventually offered, earlier intervention might have helped our family cope with the emotional stress we each secretly carried. While, at this stage, the disease itself is incurable, the effect it has on the mental health of the patient is a treatable aspect of the condition—yet it receives little attention in the MD literature and, consequently, in the patient treatment.

In my current role as a Licensed Professional Clinical Counselor, Supervisor, and Educator, I intend to advocate the need for counselors as participants in an interdisciplinary approach to chronic illness management. This study marks the beginning of that journey.

Since emerging adulthood has the highest incidence rate of mental and emotional disorders compared to any other adult age group (Jonas, Brody, Roper, & Narrow, 2003; Kessler & Walters, 1998; Klerman & Weissman, 1989), and many forms of MD are diagnosed in late childhood and adolescence (Emery, 2002), it seemed important to explore how the disease and its effects are perceived by patients in this time in life. I was interested in examining this specific MD population—one that lives with the diagnosis of MD in emerging adulthood, but shows mild symptoms that will eventually progress into severe disability. I further wanted to determine how counselors could support this population emotionally, socially, and vocationally.

The adult MD literature consists mainly of quantitative studies written for medical doctors and genetic counselors, which does little to help others understand the experience of emerging adults living *with* the disease. What little exists almost exclusively focuses on the Duchenne type of MD which is typically diagnosed in childhood (by age 5). It offers virtually no insight in the experience of emerging adults with different forms of the disease. It does highlight that those with MD have a lower quality of life. Results are mixed regarding how much of that is related to severity of disability. Inadequate coping mechanisms (Nätterlund & Ahlström, 1999a); difficult emotions (Nätterlund, Sjöden, & Ahlström, 2001), and mental strain caused by the disease (Aho, Hultsjö, & Hjelm, 2016)

have been identified. The existing literature does not explain how and when these feelings and processes begin, and what may improve the otherwise dim physical and psychosocial outcomes in adulthood.

The descriptive phenomenological method of inquiry was an ideal fit for this study. This empirical approach gathers comprehensive descriptions from emerging adults living with JOMD that served as the data for analysis and synthesis to reveal the essential structure of the phenomenon under investigation (Moustakas, 1994). Phenomenologists suspend, or bracket, their assumptions while gaining an interpretive understanding of the subjective experience of the participant (Giorgi, 2009). The empirical approach examines how individuals with a shared experience perceive it in consciousness (Giorgi, 2009). In other words, how do emerging adults experience and describe their illness and progression psychologically, regardless of the degree of actual physical severity. This study is the first to examine the narratives of emerging adults living with JOMD.

Statement of the Problem, Purpose, and Rationale

The mental health needs of people with MD are virtually overlooked in clinical practice and empirical literature. Early intervention and treatment of mental and emotional health has been associated with better disease management for such conditions as diabetes (Browne, Nefs, Poutwer, & Speight, 2015), systemic sclerosis (Kwakkenbos et al., 2014), and asthma (Maslow, Haydon, Ford, et al., 2011). A need for such services in the MD population has been identified (Ahlström & Wenneberg, 2002; Reid & Renwick, 2001), but no further information regarding the extent and nature of such

interventions is described. While the existing literature points to issues such as lower quality of life (Padua et al., 2009), emotion focused coping (Nätterlund & Ahlström, 1999a) and emotional distress (Boström & Ahlström, 2004; Nätterlund et al., 2001), much of the information is gathered on pre-constructed quantitative assessments that do not consider the context of the disease severity, type, or developmental influence.

Studies of the human lifespan suggest that emerging adulthood is a period of high stress, low stability, and increased probability to be diagnosed with a mental or emotional disorder (Ge, Conger, & Elder, 2001; Koenig & Gladstone, 1998). Research suggests that emerging adults living with a chronic illness since childhood are at an even higher risk for developing psychopathology (Bernstein et al., 2013; Kiviruusu et al., 2007) and experiencing poor social outcomes (Kokkonen, Kokkonen, & Moilanen, 2001), but results are mixed. Few focused on progressive, degenerative, non-curable disorders such as MD.

What happens during the transition from adolescence to adulthood has great impact on young people's futures. Common developmental milestones encountered in emerging adulthood include finishing education, starting a career, engaging in long-term partnerships, and deciding to have children (Arnett, 2015; Setterstein & Ray, 2010). No literature examining how individuals with MD navigate these milestones exists. The paucity of existing research warranted an exploratory study to begin to understand what it means to live with JOMD in emerging adulthood.

Thus, the purpose of this study was to understand how emerging adults perceive and describe their experiences of living with JOMD. It is assumed that there is an

"essential structure" among this population that transcends the physical symptoms. Illuminating the lived experiences of this population will help mental health professionals, medical doctors, and family members better understand and treat the disease. Understanding the illness experience among emerging adults with JOMD could also help to normalize the experiences of others living with the condition and expedite service delivery. Such a model of care has been developed for Duchenne MD (Bushby et al., 2010a, 2010b), and this study hopes to expand and promote that initiative.

This study also adds to the literature on adolescent and emerging adulthood theory by exploring how JOMD impacts development. Each participant was approached through the lens of Arnett's theory that emerging adulthood is a developmentally and demographically distinct period in the lifespan, beginning at adolescence completion and ending with the onset of stable roles in love and work (Arnett, 2015). The research explores how those with JOMD are similar to and different from typical emerging adults.

Research Questions

Utilizing a phenomenological research design, this study regarded the illness experience of emerging adults living with JOMD as the phenomenon under investigation. Thus, the research was guided by the following questions: "How do emerging adults perceive and describe their experience of living with JOMD, beginning with their first memories of the disease up until now?" and "What do emerging adults living with JOMD believe could be helpful in adjusting to their illness?"

Definition of Terms

The following terms are relevant to understanding the study, and are defined for the reader.

Chronic disease: A condition that is non-contagious, slow to progress, and long in duration (i.e., lasts three months or more; CDC, 2018; Institute of Medicine, 2012). These conditions rarely resolve spontaneously, thus leaving the individual with limited functioning or disability, requiring rehabilitation, supervision, or long-term care (Timmreck, 1987).

The term "disease" refers to changes in the structure and/or function of body systems from "normal" to "abnormal" (Falvo, 2009). It is derived from the medical model, which focuses exclusively on treating and "curing" the pathology (Falvo, 2009; Smart, 2009). Advantages of the medical model include developing informative statistics and providing common terminology for health care professionals. Disadvantages are that it neglects the personal, social, and cultural aspects that influence the disease (Lubkin & Larsen, 2013).

Chronic illness: The perceptual response to a chronic disease (Lubkin & Larsen, 2013). Personal and psychological aspects (e.g., the experiences of disease symptoms and perceived suffering) versus the pathophysiology of the condition. Understanding one's illness experience includes examining individual, family, community and healthcare providers' responses to the disease (Lubkin & Larsen, 2013).

Emerging adulthood: The transitional period between the end of adolescence and the beginning of adulthood. It is a distinct developmental phase of life marked by five

defining features: (a) Identity exploration (especially in love and work), (b) instability (in love, work, and residency), (c) self-focus, (d) feeling in-between adolescence and adulthood, and (e) possibilities/optimism that dreams will transform into reality (Arnett, 2004). Transitions that commonly occur during this developmental period include completing school, establishing a career, financial independence, marriage (or long-term partnership), and parenthood (Arnett, 2000). Typically, emerging adulthood is defined as between the ages of 18 to 25; however, the age at which one leaves emerging adulthood and enters adulthood is highly variable, and can extend up to age 29 (Arnett, 2000, 2004). For the purposes of this dissertation, the period of emerging adulthood is defined as the period of life between 18 and 29 years of age.

Muscular Dystrophy (MD): A group of inherited diseases in which the genes that control muscle function are defective (Emery, 2002). Its defining feature is progressive and degenerative weakness of the skeletal muscles (Shannon, 2004). Some forms of muscular dystrophy affect the heart, gastrointestinal system, endocrine glands, respiratory function, and eyes (Shannon, 2004). Currently there is no treatment to stop or reverse the progression of the disease (Shannon, 2004).

Juvenile Onset Muscular Dystrophy (JOMD): A grouping of types of muscular dystrophies that typically have an onset between 9 and 18 years old, namely Becker MD, LGMD, Facioscapulohumeral MD, and EDMD (Emery, 2002). Though each of these types can be diagnosed earlier in childhood or into adulthood (Emery, 2002), the focus of this study was on the experiences of those diagnosed with a form of MD between the

ages of 9 and 18. JOMD may also be referred to as proximal MD, because the muscles most profoundly affected are located closest to the truck of the body (Emery, 2002).

Review of the Literature

This study investigates the experiences of emerging adults living with JOMD; thus, it is important to gather relevant information as groundwork. First, available research about juvenile onset chronic illness and MD is discovered and listed. Next, brief reviews of the theoretical underpinnings of adolescence and emerging adulthood are presented to situate the reader to aspects of lifespan development. Various theories of adaption to chronic illness and disability are also discussed.

Understanding JOMD

The muscular dystrophies represent a group of childhood or adolescent onset diseases that are among the least prevalent but most devastating among them all, since they are most burdensome to quality of life (Craig, Hartman, Owens, & Brown, 2016). Limited information exists describing psychological experiences of emerging adults living with types of MD that are even more rare than Duchenne, such as LGMD and EDMD, which are distinct in that individuals essentially develop normally from birth until childhood or adolescence, then decline significantly over the next decade in life. To understand how the condition is experienced, literature on pediatric onset chronic illness and MD is presented.

Pediatric chronic illness. Pediatric chronic illness is a health condition lasting at least 3 months and interferes with a child's ability to function normally, either because of physical limitations or the need for hospitalization and/or more extensive medical

attention (Secinti, Thompson, Richards, & Gaysina, 2017). A wide range of disorders could be diagnosed during this developmental period, including conditions such as asthma, arthritis, cancer, diabetes, heart disease, cystic fibrosis, and MD (CDC, 2018). Each year in the United States approximately 500,000 adolescents diagnosed with a juvenile onset chronic illness turn 18 years old, and that number is on the rise (Blomquist, 2006; Perrin, Bloom, & Gortmaker, 2007). The challenges of life as an emerging adult, such as becoming financially independent from parents, deciding on and establishing a career, exploring identity and romantic relationships (Arnett, 2000, 2015) are typically deliberated during this developmental period. Adolescents with juvenile onset chronic illness must cope at the same time with impaired health and physical functioning (Blum, 2005). Medical advancements have facilitated an increased survival rate of pediatric chronic illnesses (Perrin et al., 2007), but most research has focused on *physical* outcomes, leaving a gap in the literature regarding the mental health of those with juvenile onset chronic illness. Thus, it has been a topic of growing empirical interest.

Psychological issues. Results from a recent meta-analysis suggest that adults who grew up with a chronic illness are more likely to experience emotional problems, particularly depression and anxiety, compared to healthy peers (Secinti et al., 2017). Theories linking mental and physical health, such as the stress-diathesis model, predict that emerging adults with juvenile onset chronic illness are at high risk for developing mental and emotional disorders. The stress-diathesis model explores the interaction of nature and nurture in the development of psychopathology. It posits that everyone has a set of genetically inherited vulnerabilities (i.e., diatheses such as depression or

schizophrenia) that, when activated by environmental stress, lead to the emergence of a disorder (Burke & Elliott, 1999; Sabharwal, 2014). Emerging adults with chronic illness are exposed to a higher allostatic load (persistent wear and tear on the body) and stress (Eddington, Mullins, Byrd-Craven & Chaney, 2012), thus increasing the risk of psychopathology (Ferro, Gorter, & Boyle, 2015). Those with chronic conditions endure periods of high stress that disrupts employment goals, economic lifestyle, and personal aspirations as symptoms are experienced, which can elicit emotional turmoil and grief (Turner & Kelly, 2000). Young adults with chronic illness desire to live a "normal" life, but at the same time must manage a potentially life-threatening condition that imposes obstacles on the affected individual, parents and siblings (Waldboth et al., 2016). According to cognitive-behavioral theories, negative thoughts, attitudes, or beliefs that exaggerate the impact and associated harm of the illness can produce symptoms of depression and anxiety (Beck, Emery, & Greenberg, 1985). Individuals who report high levels of learned helplessness and focus on the negative aspects of their condition have shown to experience worse psychological functioning, even after adjusting the results for disease severity (Evers et al., 2001; Nieto, Raichle, Jensen, & Miró, 2012).

Recent empirical studies have examined the rate of mental and emotional disorders among pediatric onset chronic disease patients. In one longitudinal study, Ferro et al. (2015) followed 2,825 youths with various chronic illnesses from ages 10–11 to 24–25 years of age to examine depressive symptoms. Compared to a "healthy" control group of age related peers, those with a chronic illness consistently experienced a higher rate of depression. Symptoms increased from early to late adolescence (ages 13–18),

declined from ages 18 to 23, then increased again in the latter half of emerging adulthood (ages 24–25). The prevalence of suicidal thoughts, plans, and attempts is also higher among adolescents and young adults with chronic illness (Ferro et al., 2017). The severity of the chronic illness and its degree of disability was found to be associated with suicide ideology partially due to the volume of negative life events experiences, which increases vulnerability to hopelessness (Zhou & Chen, 2017). Ferro et al. (2017) postulated that social isolation and dependency on others caused by the chronic illness impacted thoughts about killing oneself. Other studies have confirmed higher rates of depression (Bernstein et al., 2013; Kiviruusu et al., 2007) and anxiety (Bernstein et al., 2013; Packham, 2004). Kokkonen et al. (2001) found similar prevalence rates of psychiatric disorders between emerging adults with chronic illness and healthy peers, but those with a chronic disease experienced more severe psychiatric symptoms that more profoundly affected everyday life.

Given that the earlier one experiences a mental or emotional disorder, the longer the disorder will last, and the greater opportunity for relapse exists (Kovacs, Feinberg, Crouse-Novak, Paulauskas, & Finkelstein, 1984), emerging adulthood is a critical period for identification, prevention, and intervention services. Furthermore, statistics indicate that mental health disorders significantly and negatively affect disease management and adherence to treatment interventions (Bernstein et al., 2013; Pai & McGrady, 2013). These studies suggest that it is not uncommon for mental illness to run alongside chronic illness, and that treating the psychiatric issues can improve health and psychosocial well-being (Verhoof, Maurice-Stam, Heymans, Evers, & Grootenhuis, 2014).

Social issues. In a meta-analysis of studies comparing the achievement of developmental milestones among young adults with and without chronic illness, Pinquart (2014) found that those with chronic illnesses were 22–23% less likely to complete higher education and leave the family home, 25% less likely to establish employment, 27% less likely to be married, and 38% less likely to have children. The results of individual studies, however, are quite mixed. Maslow and colleagues (Maslow, Haydon, Ford, & Halpern, 2011; Maslow, Haydon, McRee, Ford, & Halpern, 2011) found that those with asthmatic chronic illnesses were just as likely as their non-disabled peers to establish committed romantic relationships, have children, and live with parents, but less likely to obtain a college degree and establish employment. Those with *non-asthmatic* illness (e.g., cancer, diabetes, epilepsy) had significantly worse outcomes on all measures. All emerging adults with juvenile onset chronic illnesses were less likely to be employed, earned a lower income, less likely to graduate college, and had higher odds of receiving government assistance (Maslow, Haydon, McRee et al., 2011). Disturbances in completing developmental tasks may threaten adult identity (Johnson, Berg, & Sirotzki, 2007), quality of life, and emotional health (Maurice-Stam, Grootenhuis, Caron, & Last, 2007).

Growth into adulthood with a juvenile onset chronic illness has also been shown to disorient social development (Kokkonen et al., 2001). A Finnish study of 407 emerging adults (aged 19 to 25 years) with chronic illness since childhood found that those with a chronic disease were almost twice as likely to have delayed social maturation, as measured by the Social Maturation Index (Kokkonen et al., 2001). Risk

factors associated with poor social maturation included being a male, residing in a rural environment, poor school achievement, lack of vocational training, poor employment status, familial conflict during childhood, and early dating (Kokkonen et al., 2001).

Healthcare issues. Literature indicates yet another social concern for emerging adults with a juvenile onset chronic illness: The transition from pediatric to adult healthcare services (Colver et al., 2013). The concept of "emerging adulthood" has empirical support for being a distinct developmental stage bridging adolescence and adulthood, eliciting feelings of being "in-between" the two stages (Arnett, 2000). It assumed a slow, gradual transition to adult responsibilities (Arnett, 2000, 2015). Unfortunately, the healthcare system has yet to be adapted to this cultural shift. Individuals with a chronic childhood illness are abruptly cut off from services when they turn 18 years old, and lose access to public support in the education, healthcare, and mental health service domains (Osgood, Foster, Flanagan & Ruth, 2005). They are expected to make the most dramatic healthcare transition in life during the most unstable, unpredictable, developmental period, and many fail to do so successfully (Rapley & Davidson, 2010). In addition to the loss of relationships with familiar, trusted professionals, the locus of control shifts from a family focus to an individual (or patient) focus (Kirk, 2008; Rapley & Davidson, 2010). The experience of transitioning from pediatric to adult healthcare services has been described as a time of uncertainty, grief, and confusion regarding the patient's role in disease management (Kirk, 2008; Rapley & Davidson, 2010). In sum, emerging adults lose familiar healthcare services when they potentially need them the most. Years may pass without any service utilization because

individuals lack the knowledge of how to find providers, believe that they do not need healthcare, or are anxious about initiating contact.

Access to affordable mental health care professionals with knowledge of or experience in chronic disease is another identified obstacle (Hunt, 2009). In a qualitative study examining psychological and social needs of adult women with various neurological diseases (i.e., multiple sclerosis, spinal bifida, spinal cord injuries), participants expressed dissatisfaction with psychologists and mental health counselors for reasons ranging from inaccessible office locations, "very bad attitudes" (p. 7) toward disabilities, and lack of understanding about their disease among service providers (Hampton, Zhu, & Ordway, 2011). Counselors themselves do not know how to treat this growing population, expressing little training in their graduate programs about chronic disease and disability (Oliver, 1995).

Nonetheless, the need for such services is clear. In Hampton et al.'s (2011) study of adult women with chronic illness, 64% of the sample was currently diagnosed with a major depressive or anxiety disorder that was pharmacologically treated. One participant stated:

> I really think that social and counseling services that target adults with spinal bifida and their needs are lacking. They had a group for SB kids and their parents when I was younger but when I became an adult, they don't have anything for me. (Hampton et al., 2011, p. 7)

Even if an emerging adult with a juvenile onset chronic illness has the motivation and insurance coverage to seek counseling services, there is a strong possibility the

professional will lack the knowledge about the condition. This points to a gap in the literature merging mental healthcare and emerging adults with chronic disease. Of the limited research that does exist, much of it focuses on common chronic diseases, such as diabetes (Browne et al., 2015), asthma (Maslow, Haydon, Ford, et al., 2011), epilepsy (Gauffin, Flensner, & Landtblom, 2011), cancer (Kumar & Schapira, 2013), and cerebral palsy (Magill-Evans, Wiart, Darrah, & Kratochvil, 2005). The research is also mainly quantitative. Much remains unknown about the psychosocial effects of degenerative (i.e., continual breakdown of the affected anatomy's structure and function) and progressive (i.e., the disease will continue to worsen and health declines are inevitable) juvenile onset chronic illnesses, such as MD, during emerging adulthood. Managing chronic illness is as much a psychosocial issue as a medical one (Smart, 2009); therefore, it is important to go beyond the medical setting to understand how emerging adults perceive and describe the mental and emotional struggles that occur alongside the disease.

Muscular Dystrophy (MD). MD is an umbrella term for genetic diseases that progressively weaken and degenerate skeletal muscles used for voluntary movement (Shannon, 2004). While the onset of MD can come later in life, 80% of individuals with the disease exhibit symptoms before adolescence (Papazian & Alfonso, 2002). MD is the most devastating muscle disease for several reasons: (a) it is a slow, insidious progression of muscle weakness that cannot be prevented, leading to (b) inevitable muscle damage and loss of functioning, causing not only a limited range of movements but breathing and feeding difficulties and possible early death; (c) lack of medicinal, pharmaceutical and surgical treatment options for symptom alleviation or remission, and (d) incurability of

the illness (Gawlik, 2018). The insidious, physically disabling, course of MD is unique and different from many other more common, stable, chronic illnesses. For example, some individuals with type 2 diabetes mellitus reported relief after diagnosis because they could prevent further damage by proactively monitoring blood glucose readings (Peel, Parry, Douglas, & Lawton, 2004). Secondary complications could be avoided. On the other hand, nothing can be done to stop or even slow down the paralysis in MD.

The forms of MD commonly diagnosed in childhood include: Congenital, Duchenne, Becker, Limb-girdle, Facioscapulohumeral and Emery-Dreifuss. Each are described in more detail in a later section, and are presented in Table 1. Different muscle groups are affected in each type. However, of the 434 voluntary muscles that can be affected, the spine and limb-girdle muscles (shoulders and hips) are the most frequently and profoundly degenerated in MD (Emery, 2002). Some types affect the heart, gastrointestinal system, endocrine glands, skin, eyes, and other organs (Shannon, 2004). However, the defining feature of all types of MD is a slow deterioration of voluntary muscle function.

Crude estimates suggest that MD affects approximately 250,000 American children and adults (Shannon, 2004). The disease is known around the world, but this researcher has not found data on prevalence outside of the USA. Population-based or specific type prevalence estimates exist but pooled data is unavailable (Gawlik, 2018; Mah et al., 2014). Duchenne MD, the most common childhood form, has an American incidence rate of about 1 in 7,250 males aged 5–24 years old (Romitti et al., 2015). Most forms of MD are rare diseases, such as LGMD Type 1a, affecting <1 per 100,000 people

(Gawlik, 2018). However, these are crude, preliminary estimates. Advances in cloning, genetic mapping, and sequencing have shed more light on the complexity of MD (Gawlik, 2018).

MD most commonly affects the muscles by destroying proteins in the muscle fiber membrane. Muscle fibers are a group of individual cells that are joined together and encapsulated by a membrane. These individual fibers are arranged into bundles, which make up skeletal muscle. When muscles are activated by an impulse from the brain and spread through the central and peripheral nervous system, the muscle fiber membrane protects the cells as they contract and relax. A group of proteins, called the dystrophin-glycoprotein complex, stabilize the muscle cell when it is under stress. In most individuals affected with MD, the dystrophin-glycoprotein complex is compromised, leading to chronic muscle damage. In MD patients, muscle fibers continually leak creatine kinase, a protein essential for muscle contractions. As creatine kinase is released, the muscle cells absorb excess calcium, which eventually causes cell death and muscle degeneration. Quercia (2012) described this process as a tent with faulty poles. The tent (muscle cell) ends up collapsing and is replaced by fat tissue.

The term “muscular dystrophy” is descriptive in nature for the many different types of MD, each of which have unique molecular breakdown patterns, physical symptoms, genetic causes, inheritance patterns, ages of onset, and levels of severity. What is common across all types of the disease is that it progressively degenerates skeletal muscle.

Table 1

Forms of Muscular Dystrophy

Type	*Onset*	*Life Expectancy*	*Limitation*	*Inheritance Pattern*
Congenital	Birth/early onset Diagnosed: 0 - 1 years old	Ranges from early death to normal lifespan	Generalized muscle weakness; mobility impairment; respiratory complications, spinal curvature; learning disabilities; mental retardation, eye defects; seizures	Affects males and females
Duchenne	Birth/early onset Diagnosed: 2 – 6 years old	Death usually occurs in late teens or early 20's	Muscle weakness most profound of all types, causing mobility impairment; respiratory issues, cardiac impairment; possibly intellectual impairment	Affects only males
Becker	Juvenile Onset Diagnosed: 5-15 years old	Without heart complications, lifespan may be normal. With cardiac involvement, lifespan may be shortened	Muscle weakness similar to Duchenne, but not as severe; mobility impairment; cardiac impairment; possible cognitive deficits	Affects only males
Limb-Girdle	Juvenile Onset Diagnosed: 10 - adulthood	Ranges from early death to normal lifespan	Muscle weakness in hips, shoulders; mobility impairment; respiratory and cardiac involvement	Affects males and females
Facioscapulohumeral	Juvenile Onset Diagnosed: by age 20	Life expectancy not shortened	Facial weakness, especially eyes and mouth; speech difficulties; weakness of shoulders, upper arms, lower legs, hip and abdominal muscles	Affects males and females
Emery-Dreifuss	Juvenile Onset Diagnosed: by age 10	Without serious cardiac complications, can survive until middle age or late adulthood	Early development of muscle contractures before muscle weakness; sudden death possible from cardiac complications; mobility impairment	Primarily affects males; can affect females

Note. Emery, 2002; Shannon, 2004

Types of pediatric MD. Of the nine different forms of MD, six of them are diagnosed before an individual reaches emerging adulthood. It is important to understand not only the different types of MD, but also how patients diagnosed in early childhood live with different physical, psychological, and social disease experiences compared to those diagnosed in later childhood. Therefore, the six types are divided into two categories, Birth/Early Onset and Juvenile Onset, based on phenotype similarities.

Birth/early onset MD. The following section describes the two forms of MD that are typically diagnosed between the ages of 0 and 5: Congenital MD (CMD) and Duchenne MD (DMD).

Congenital MD (CMD). CMD is a group of dystrophies that are present at birth, causing muscle weakness within the first few months of life (Emery, 2002). Newborns typically have hypotonia (floppiness), poor head control, and a delay in meeting other developmental motor milestones (Shannon, 2004). Unlike all other forms of MD, the muscle weakness present in babies with CMD is not necessarily progressive and degenerative (Emery, 2002). Some children even learn to walk (Emery, 2002). Nonetheless, contractures often develop due to immobility (Emery, 2002). Shannon (2004) identified three main types of CMD: (a) Those with symptoms of muscle weakness; (b) children with both muscle weakness, mild to severe learning difficulties, and possible epilepsy; and (c) those with muscle weakness, mild to severe learning disabilities, and abnormalities of the eye. Heart and respiratory complications may also develop (Emery, 2002). It has an autosomal recessive inheritance pattern (Emery, 2002). The lifespan is likely to be shortened due to complications from CMD (Gawlik, 2018).

Duchenne MD (DMD). DMD is the most prevalent of all forms of MD, with an incidence rate of 1 in 5000 live male births (Wicklund, 2013). Females can be genetic "carriers" of the disorder, but do not display symptoms nearly as dramatically as males (Wicklund, 2013). The average age of diagnosis is age 5, though boys begin to show weakness as early as 3 years old (Shannon, 2004). Toddlers with DMD are often late in learning how to walk. Caregivers may notice enlarged calf muscles (i.e., pseudohypertrophy). As the child approaches preschool years, he may seem clumsy and fall frequently. Soon, other noticeable symptoms include difficulty rising from a chair or the floor, climbing stairs, and maintaining balance (Shannon, 2004). The disease gradually affects the back, shoulders, trunk, and the upper and lower legs. By school age, the patient has a waddling gait and falls easily. To improve balance, children with DMD will stick their stomachs out and pull their shoulders back (Shannon, 2004). It also becomes difficult to raise their arms over their head. Most children lose the agility to walk between the ages of 7 and 12, and more than 90% of males require a wheelchair for mobility by the time they reach 15 years old. During adolescence, boys with DMD experience the most significant loss of skeletal muscle strength (Shannon, 2004). The heart and respiratory muscles become compromised during late adolescence and/or early adulthood (Shannon, 2004). The myocardium in the heart begins to deteriorate (similar to the skeletal muscles), increasing the risk of heart attack or respiratory failure (Shannon, 2004). Unfortunately, many males die from these complications by the time they reach 25 years old (Shannon, 2004). The mean lifespan for DMD patients has increased over the last decade, thanks to improvements in care and corticosteroid use,

from 19 years to more than 25 years (Wicklund, 2013). There is an increased need for physical and psychosocial support as these individuals transition from childhood into adulthood.

Juvenile onset MD (JOMD). The term "Juvenile Onset MD" was created to incorporate forms of MD diagnosed in late childhood or adolescence, or approximately between the ages of 9 and 18 (not to suggest one cannot be diagnosed as an adult, but that was not the focus of this study). These types of MD have also been referred to as the "Proximal" MD group, because the muscles most profoundly affected are located closest to the trunk of the body (Nätterlund et al., 2001). Emery (2002) identified four types of JOMD, which include: Becker MD, EDMD, LGMD and Facioscapulohumeral MD.

Becker MD (BMD). BMD affects approximately 3 out of every 100,000 live male births (Shannon, 2004). Female carriers may show mild symptoms of the disorder, but like DMD, cannot have the disorder (Wicklund, 2013). The average age of diagnosis is 12 years old (Emery, 2002). Symptoms begin in childhood, with muscle cramps and poor athletic performance being the first noticeable indicators. Trouble walking, running, and climbing stairs become evident in the teens and twenties. Loss of ambulation varies from adolescence onwards (Wicklund, 2013). BMD also affects the muscles in the shoulders, upper arms and neck, though not as dramatically as the lower extremities (Shannon, 2004). The heart and respiratory muscles may be compromised as well (Shannon, 2004). Death usually occurs in the fourth or fifth decade of life, making BMD the severest form of JOMD (Emery, 2002). There is a high recurrence rate, or possibility

of passing on the gene to future generations, because all daughters in a family with an affected son will be carriers (Eggers & Zatz, 1998).

Limb-girdle MD (LGMD). LGMD is a heterogeneous group of rare, genetically inherited disorders, with an estimated prevalence varying from 8.1-69 per million people (Rosales & Tsao, 2012). It predominantly affects the muscles in the hip and shoulder regions, also known as the pelvic and shoulder girdles, which collectively are known as the limb girdles (Shannon, 2004). LGMD can be classified into two major categories, autosomal dominant (i.e., LGMD type 1) and autosomal recessive (LGMD type 2; Rosales & Tsao, 2012). The researcher of this study (Kelsey Jager) is affected with LGMD (type 2a). Muscles attached to the limb girdles, such as the thighs, shoulders, chest, and biceps, are most impacted, causing initial difficulties such as climbing, running, and getting up from the floor (Rosales & Tsao, 2012). Occasionally, there is cardiac and respiratory involvement (Shannon, 2004). Over the last four decades, researchers have identified over 20 different subtypes of LGMD, making this disease one of the most complex, evolving, and elusive forms of MD (Rosales & Tsao, 2010). The different types of LGMD each have unique causes that lead to muscle weakness. Bushby (1999) provided a comprehensive description of the various types and the specific molecular genetics involved, which are beyond the scope of this review. Suffice it to note that different types of LGMD have contrasting ages of onset, varying degrees of severity, and inheritance patterns (Shannon, 2004). Individuals afflicted with LGMD typically lose the ability to walk within 20 years of diagnosis, though progression varies

(Shannon, 2004). Like other forms of MD, there is currently no treatment to stop or slow the progression of the disease (Shannon, 2004).

Facioscapulohumeral MD (FSHD). FSHD is the third most common form of MD, affecting approximately 1 in 20,000 individuals (Statland & Tawil, 2014). It is a dominantly inherited disorder that primarily affects the muscles of the face, shoulder blades, and upper arms, though other muscles are also degenerated (Shannon, 2004). Individuals with FSHD typically display symptoms before their 20th birthday, beginning with mild facial weakness and subsequent scapular involvement (Bakker, Schipper, Geurts, & Abma, 2017). Difficulty reaching the arms above shoulder level is often the first noticeable symptom (Statland & Tawil, 2014). Lower-extremity weakness ensues as the disease progresses, leaving some individuals to become wheelchair bound (Statland & Tawil, 2014). However, like in other forms of MD, the severity of FSHD varies. While some patients are mobility impaired, others can be nearly asymptomatic (Statland & Tawil, 2014). Other issues that can occur with the disease include hearing and vision loss, respiratory weakness, and cardiac involvement (Statland & Tawil, 2014). The progression is usually slow, and individuals with FSHD can expect a normal life span (Statland & Tawil, 2014).

Emery-Dreifuss MD (EDMD). People with EDMD are usually diagnosed around the age of 10 years old, but the age of onset is variable, from childhood into adulthood (Gawlik, 2018). The disease can be inherited as a sex-linked trait like DMD and BMD, or as an autosomal dominant trait, which affects both males and females (Shannon, 2004). Three hallmark characteristics make this disease unique. First, there are

contractures, or tightening and shortening of muscle groups, causing stiffness at the joints, *before* muscle degeneration (Emery, 2002). Typically, contractures develop at later stages of muscle wasting conditions due to inactivity, making this event a distinctive phenomenon in EDMD (Shannon, 2004). The Achilles tendons, elbows, neck, and spine are affected (Emery, 2002). Second, the muscle weakness has a unique pattern that manifests itself mainly in the upper limbs (i.e., shoulders and upper arms) and proximal lower limbs (i.e., calf muscles of the leg; Emery, 2002). In late stages of the disease, the limb-girdle musculature is weakened, making it difficult to climb stairs and rise from a sitting position (Shannon, 2004). Third, the heart is affected in a way that is unusual for individuals with MD; rather than weakening the heart as a muscle, it compromises the cardiac conduction system that controls the rate at which the heart beats (Shannon, 2004). Issues include abnormally slow heart rate (sinus bradycardia), alterations in the rhythm of the heartbeat (prolongation of the PR interval), and complete blockage of the electrical signal that stimulates the heartbeat (Emery, 2002). Cardiac involvement can occur without skeletal muscle weakness. Because the symptoms can go undetected, EDMD can cause sudden death in seemingly healthy emerging adults. In other individuals, cardiac disease occurs simultaneous with severe muscle weakness (Emery, 2002). A pacemaker or defibrillator can be utilized to ensure the heart functions normally (Shannon, 2004). Though the lifespan may be shortened, many individuals reach middle age and beyond.

These types of JOMD display similar phenotypes in progression pattern and, in some cases, severity. Apart from LGMD, nearly all individuals with BMD, FMD and EDMD are diagnosed between the ages of nine to 16 years old, though diagnosis can

occur later in life (Emery, 2008). This "group" of disorders has a slow progression during adolescence or early adulthood (Aho, Hultsjö & Hjelm, 2015). The earlier the symptoms are first noticed, the more severe the disease (Gawlik, 2018). While those with JOMD can expect a longer period of mobility and independence, it also requires adaption as muscle loss becomes more apparent in emerging adulthood, eventually and inevitably forcing them to request support from others to manage activities (Aho et al., 2015). To demonstrate the difference between those with birth/early onset MD and JOMD, emerging adults with DMD are severely disabled and considering end of life planning while those with JOMD may be just beginning to show mild symptoms.

Emerging Adults Living With JOMD

Emerging adults with JOMD are at an important juncture in life, because they are making decisions that promote the transition from adolescence to adulthood while their condition is becoming more noticeable. The insidious, slow, and debilitating nature of JOMD makes it unique from a lifespan perspective. Emerging adults living with JOMD may experience the disease more psychologically than physically, as symptoms are typically mild in the first few years of onset. As the research on chronic illness has blossomed, healthcare providers now know that how an individual interprets and makes meaning of the illness plays an important role in the adjustment process (Evers et al., 2001; Nieto et al., 2012). The experiences and decisions of this developmental period can have a powerful impact on adulthood; how emerging adults perceive and experience their disease will impact how they cope, their utilization of services, their identity, and belief in their ability to accomplish developmental milestones.

Healthcare providers overlook emerging adults with JOMD (Aho et al., 2016). With no available treatment options and minimal physical symptoms compared to their birth/early onset MD counterparts, they may appear to be minimally affected by the disease. In other words, these individuals may appear healthy, thus having a more difficult time acquiring or accepting assistance. Psychologically, though, emerging adults with JOMD might be at the most difficult adjustment process of their life. They are possibly coming to terms with the diagnosis, learning more about the disease, experiencing physical progression, and trying to plan for the future when the future is a frightening place. One individual (age and type of MD unknown) described it as follows:

> I feel I'm a damned hindrance to myself, mostly mentally . . . But anyway I don't feel secure, I'm so afraid. I don't know what's going to happen when I get worse—and I can see that I am getting worse, even though it's a slow process. I'm scared to death because I know I'm getting worse—I exercise a lot and try to fight it, but there's no stopping it. It's like a bulldozer flattening me. But I've had too much of a yearning to be healthy, I must admit—far too much for my own good, because it makes you feel bad in the end. (Boström & Ahlström, 2004, p. 1392)

Emerging adulthood may be an ideal time for mental health services, because it is in emerging adulthood that patients with JOMD begin to show more noticeable symptoms, thus increasing stress and the likelihood of developing mental and emotional disorders. By uncovering the lived experiences of emerging adults with JOMD, medical and mental health professionals can more comprehensively treat the disorder. Currently,

it is unclear how, and to what extent, JOMD impacts emerging adulthood, and if there are interventions that might improve the, otherwise poor, psychological and social prognosis. This study seeks to add to the MD and emerging adulthood literature by providing insight into how emerging adults affected with JOMD experience the illness.

Only three studies were located that examine the experiences of emerging adults living with JOMD, and all were written by Aho, Hultsjö and Hjelm (2015, 2016, 2017). Individuals between the ages of 20 and 30 living with LGMD Type 2a, a rare, recessive form of the disease served as the sample. Utilizing the salutogenic orientation as a theoretical framework, these researchers identified themes in the comprehensibility (cognitive category referring to how one understands and interprets the disease), manageability (behavioral category focused on coping mechanisms activated in response to the disease) and meaningfulness (motivational category identifying experiences that bring personal happiness and satisfaction) domains. The comprehensibility category showed that young adults with LGMD2a and their parents experience both physical and emotional duress, especially in reaction to the continual adjustments required as the diseases progresses (Aho et al., 2015, 2017). One participant in one of these studies stated, "I suppose that's what I think is the major thing about the diagnosis, that it's constant change . . . adapting all the time to the body and new situations" (Aho et al., 2015, p. 4). Transitioning from ambulatory independence to a wheelchair was described as psychologically distressing. Anxiety about one's health in the future was another theme in the comprehensibility domain. Parents reported repressing feelings related to

unresolved problems (Aho et al., 2017). Participants described negative thoughts dealt with by focusing on the present. One participant described her experience as follows:

> Now I'm alive and this is how I feel . . . if I could just think, "OK, it's fine now" and not think so much about the future, that it can get worse, then everything would be good, but there are those thoughts that keep spinning, "What will happen?" . . . you have to take one day at a time. (Aho et al., 2017, p. 5)

In a later publication, Aho et al. (2016) described health perceptions related to sense of coherence, or the ability to comprehend one's health situation and the capacity to make use of available resources along an ease/dis-ease continuum (Antonovsky, 1993), and found that mental strain, negative attitudes from society, and accessibility problems interfered with movement towards the healthy pole.

In the manageability domain, Aho et al. (2015) highlighted the difficulties emerging adults with LGMD2a face in accessing external sources of professional support services such as medical checkups, assistive devices, financial support, and home adaptations, to name a few. Negative emotions were managed by leaning on friends and family members. Parents are found to be an important source of support and caregiving for those with LGMD, in addition to a strong social network, contact with well-liked professionals, and personal care assistance (Aho et al., 2017). Time spent caregiving puts parents' work satisfaction and well-being at risk (Hilbrecht, Lero, Schryer, Mock, & Smale, 2017). Emerging adults with LGMD expressed independent efforts to maintain mental well-being by positive thinking and hopeful thoughts about the future, and an interest in professional counseling.

In the final domain, meaningfulness, Aho et al. (2015) reported that emerging adults with LGMD 2a experience joy from identity and sense of purpose that comes with employment and socializing with friends. Fatigue impeded engagement in meaningful activities, and resulted in a range of negative experiences, such as inability to participate in sports previously enjoyed, reduction in daily activities completed, trouble keeping up with occupational and personal demands, and having to leave work altogether (Aho et al., 2015). Parents hoped their child with LGMD would live a fulfilled life, defined as participation in and support from the community, perceived happiness, personal independence, and engagement in meaningful and personally interesting activities (Aho et al., 2017). Overall, these findings indicate that the young adults in this study had trouble managing daily life; the services that participants did receive were frustratingly difficult to acquire and were preceded by continual explanation and justification of needs.

Aho et al.'s (2015, 2016, 2017) studies were the first to address the gap in the research by describing, through the lens of the salutogenic orientation theory, how one group of Swedish young adults with LGMD Type 2a experience the disease. Much remains unknown about how JOMD is experienced, so the next section broadens the search to include research investigating all forms of MD across the adult (versus emerging adult) population.

Psychosocial Consequences of Adult MD

The literature examining psychological and social aspects of MD is scarce. Most research has been dedicated to curing the disease, thus it is written by and for medical professionals and geneticists. Of the limited psychosocial research, much of it focuses

exclusively on the Duchenne type, likely due to the high prevalence rate and severity of the disease. Outside of the medical and DMD literature, there is a mixed body of research among the adult MD population (Krause-Bachand & Koopman, 2008). This section serves to highlight important themes related to this study, but is not meant to be all inclusive.

Researchers have identified psychosocial issues and needs of emerging adults with DMD, which include the presence of a personal care aide, problems with education and employment, difficulty maintaining independence, social isolation, and dealing with a terminal illness (Kinnett et al., 2007). Interestingly, the rate of depression in boys with DMD is highest during childhood, or between 8 and 11 years old, compared to adolescence or young adulthood (Elsenbruch, Schmid, Lutz, Geers & Schara, 2013). Existing research on emerging adults with DMD is promoting a "normal" transition to adulthood by encouraging independence, higher education, and career planning, while providing opportunities to discuss the presence of the disease in terms of physical deterioration, pain, and the emotional impact of medical interventions and impending death (Abbott & Carpenter, 2015).

The level of family stress is an important factor contributing to the psychosocial adjustment, especially among patients with MD (Aho et al., 2017; Bowen, 1976; Reid & Renwick, 2001). A grieving period typically follows news of a child's diagnosis: shock, denial (that the diagnosis is correct), anger, blaming, fear, and acceptance (George, Vickers, Wilkes, & Barton, 2007; Graungaard & Skov, 2007; Wright, 2008). Mothers are particularly prone to exhibit great stress, especially after learning of the hereditary

nature of the disease by which they themselves are unaffected (S. P. Harris, Heller, & Schindler, 2012). Witte (1985) found that parents of 13- to 16-year-old boys felt guilt, shame, sorrow, anger, and fear when discussing their child's disease. The task of explaining the disease or answering questions falls on the caregiver, as the affected child ages and becomes more aware of the physical disability (Emery, 2002).

Mothers and fathers of children with birth/early childhood onset and juvenile onset muscular dystrophy often serve multiple roles of parent, caregiver and medical advocate while working full-time (George, Vickers, Wilkes & Barton, 2008; Waldboth et al., 2016). The physical, mental and emotional demands of caregiving lower perception of well-being (George et al., 2008; Hilbrecht et al., 2017). A recent investigation of parents of young adults with LGMD2 reported the disease having a major impact on life not only by caregiving duties but also difficulty understanding symptoms and worries about the future (Aho et al., 2017).

For those with JOMD, major disease progression occurs at the crossroads of adolescence and emerging adulthood. Psychosocial intervention services for DMD throughout the course of the disease and over the lifespan of a person with DMD have been developed (Birnkrant et al., 2018; Bushby et al., 2010a, 2010b) but only recently have researchers begun to assess the experiences of those diagnosed with different types of MD, such as LGMD, indicating that more individual and family support is needed, especially during the transition into a power wheelchair (Aho et al., 2018).

Excluding DMD, research among the adult population indicates MD has a significant impact on the life of the affected individual, their family members, loved ones,

and caregivers. Learning of one's diagnosis has been described as traumatic (Nätterlund et al., 2001). Adults with MD and cancer were compared regarding the reaction to being diagnosed and coping abilities, and Ahlstrom and Sjödén (1994) found an 82% overlap. New categories classifying the MD experience were anticipation, minimization, secretiveness, fear, social comparison, establishment of control over everyday life, and creation of new life values (Ahlstrom & Sjödén, 1994). It can be concluded that being diagnosed with MD is a stressful, emotionally difficult life event, yet few follow-up procedures exist.

The progressive nature of MD leads to repeated losses of abilities perceived as valuable (Nätterlund et al., 2001). This process has been described in the literature as chronic sorrow, or stressful periods followed by times of happiness or constancy (Nätterlund et al., 2001). Losses often experienced by persons with MD include independence, control, status, social roles, and one's identity as a healthy person (Nätterlund et al., 2001). One lives in a state of awareness that health declines will occur (e.g., losing the ability to walk) but not knowing when and how they will manifest themselves. Among emerging adults with LGMD2a, the concept of loss is described as continual adjustments to slow muscle weakening that, at times, created psychological distress and depression (Aho et al., 2015). It is unclear to what degree loss impacts those with JOMD.

Nätterlund and Ahlström (1999a) showed individuals with MD bestow emotion-focused coping strategies, or attempting reduce negative and distressing feelings associated with stress rather than engage in problem-focused coping, in which an

individual reduces stress by going to the source, or addressing the problem itself (Lazarus, 1993). Because of the perceived lack of control and unresolvable situation (i.e., incurable disease), anxiety, helplessness, and hopelessness feelings ensue (Nätterlund & Ahlström, 1999a; Nätterlund et al., 2001). Detachment, or distancing oneself from the stressful event (e.g., having MD), is another manner of emotion-focused coping (Ahlström & Wenneberg, 2002). Seeking social support is rare; individuals with MD have been shown to fear dependency on others (Ahlström & Wenneberg, 2002). Repeated losses and a shrinking social network are risk factors for emotional, social, and physical health declines.

Psychological consequences of MD described in the adult literature include emotions like anger, irritation, depression, anxiety, and panic (Boström & Ahlström, 2004; Nätterlund et al., 2001; Peric et al., 2018). Self-deprecation and shame were reported in response to perceived stigma when the disease became more noticeable (Bakker et al., 2017; Boström & Ahlström, 2004; Nätterlund et al., 2001). In a cross-sectional study of 600 adults with neuromuscular diseases in the Netherlands, self-stigma, or shame and fear about having the condition, was a stronger predictor for poorer quality of life than enacted stigma, for example, actual discrimination (Van der Beek, Bos, Middel, & Wynia, 2013). An adult with MD (age and type not indicated) described his struggle with self-esteem:

> But then it comes over you that you're worthless, and you feel worthless and it makes you unhappy. If I had this muscular disease and accepted it for what it is, it wouldn't cause me such a lot of suffering. (Nätterlund et al., 2001, p. 791)

A Tunisian adult LGMD population also demonstrated internalized blame, low self-esteem, and sadness (Miladi, Bourguignon, & Hentati, 1999). A group of Dutch researchers conducted a qualitative study exploring the illness experience of 25 adults with FSHMD, and identified intra-individual and extra-individual factors that accompany functional limitations, including fatigue, reaction to diagnosis, assistive devices, and maintaining work (Bakker et al., 2017).

Overall, studies consistently confirm that MD has a negative effect on quality of life in the mental, physical, and emotional domains (Dany et al., 2017; Padua et al., 2009; Peric et al., 2018). Yet, studies have also found a weak to moderate correlation between increased physical deterioration and worse quality of life (Boström, Nätterlund, & Ahlström, 2005; Nätterlund, Gunnarsson, & Ahlström, 2000), indicating that physical disability can only partially explain lower quality of life. Findings from focus groups among adults with slowly-progressing diseases demonstrated the psycho-emotional, social and environmental stress were the hardest aspects of the condition, over physical limitations (Dany et al., 2017). Adults with MD reported more negative disease consequences than those with post-polio syndrome, and less than half of the MD sample had arrived at "complete acceptance" of the condition despite living with it for, on average, 20 years (Nätterlund et al., 2001). Symptoms of severe fatigue and pain were reported among 54% of patients with oculopharyngeal MD, while mental health was the same as found in healthy adults (van der Sluijs, Knoop, Bleijenberg, van Engelen, & Voermans, 2016).

It is noteworthy that, of the studies that have been done concerning the psychosocial aspects of adults with MD, very few are conducted in the United States. Researchers in Sweden have studied MD as it relates to coping, and piloted a rehabilitation program (Ahlström & Sjödén, 1994; Ahlström & Wenneberg, 2002; Ahlström, Lindvall, Wenneberg, & Gunnarsson, 2006; Nätterlund & Ahlström, 1999b; Nätterlund et al., 2000; Nätterlund et al., 2001; Winblad, Lindberg, & Hansen, 2005; Winblad, Jensen, Månsson, Samuelsson, & Lindberg, 2010); investigators in Germany have researched health-related quality of life (Winter et al., 2010), obstetric issues (Rudnik-Schöneborn, Glauner, Röhrig, & Zerres, 1997), and psychiatric disorders among MD patients (Kalkman, Schillings, Zwarts, van Engelen, & Bleijenberg, 2006); researchers in the United Kingdom (Faulkner & Kingston, 1998) and Italy (Padua et al., 2009) have also contributed to the literature. Unique cultural aspects of American citizens with MD are underrepresented.

Also, most of the existing research examining the adult MD population is quantitative, providing minimal insight into what it means to live with the disease. Of the limited research that has investigated the psychosocial consequences of MD, most takes place among the adult population with heterogeneous samples of types (including forms not mentioned in this literature review that are diagnosed in later life). Most studies focus exclusively on one form of MD (e.g., LGMD) or lump together many types with a wide range of phenotype expression.

Information about emerging adults affected with other forms of MD also diagnosed in childhood is missing from the current empirical literature.

Lifespan Development

A comprehensive understanding of adolescent and emerging adulthood development is important when studying and describing the experiences of those with JOMD.

Adolescence. Adolescence—the developmental stage beginning during the teenage years and ending at age 18—is a time of considerable biological, cognitive, emotional, psychological, and social growth and change.

Physical changes. A myriad of physical growth and hormonal changes occur during this developmental period, marked by the onset of puberty. At its core, puberty transforms the body of a child into a reproductively able adult, creating visible as well as internal hormonal changes (Feldman, 2011). Sex hormones (androgens for males and estrogen for females) interact with growth hormones to cause growth spurts and puberty (Feldman, 2011). Primary sex characteristics, such as changes in the uterus, are those that directly relate to reproductive organs while secondary sex characteristics are the visible signs of maturity, like underarm hair (Feldman, 2011). One hundred years ago, girls typically began menstruation at age 14 or 15, but now experience it at an earlier age of 11 or 12, and sometimes as early as age 7 (M. A. Harris, Prior & Koehoorn, 2008). Reduced disease and improved nutrition have influenced physical growth and sexual maturation. The timing of puberty has social consequences as well. For boys, early maturation is mostly a positive experience and is associated with enhanced self-concept, popularity, and athletic success (Feldman, 2011). Early development for girls, on the other hand, may cause ridicule and involvement in sexual situations before psychological

readiness (Feldman, 2011). Late maturation poses challenges as well, where the case for girls is far better than boys. While in the short-term girls may get less attention from the opposite sex, they end up with fewer emotional problems and higher satisfaction with their slender body type (Feldman, 2011). Light and small boys are viewed as less attractive and are physically disadvantaged in athletics, leading to a decline in self-concept (Feldman, 2011). The context in which puberty occurs influences development. Though timing is one factor that impacts maturation, other factors, such as changes in peer groups, family dynamics, and school determine behavior and psychological adjustment (Mendle, Turkheimer, & Emery, 2007).

Cognitive and emotional changes. Adolescents are thinking beings, attempting to make sense of what is happening to them and what to do as they rapidly encounter new situations, people and challenges (Kuhn, 2009). The gradual cognitive advances that characterize adolescence are gradual improvements in the ability to grasp abstract concepts, think critically, and remember a vast amount of material (Inhelder & Piaget, 1958). Metacognition, or the knowledge that one has about one's own cognitive processes and the ability to monitor thoughts, is an important gain that impacts emotional development (Feldman, 2011). Teenagers are able to mentally construct elaborate scenarios about themselves and their futures, and susceptible to the perception that an imaginary audience is constantly observing their behavior (Alberts, Elkind, & Ginsberg, 2007). These changes have important implications in parental, peer, and authority figure relationships, because the egocentrism creates a self-focused worldview which elicits a high sensitivity to criticism and judgement of others (Alberts et al., 2007). Discord may

result as adolescents are able to see holes in others' explanations and question perceived imperfections in others, school, and societal ideals (Feldman, 2011).

As cognitive growth occurs, so does emotional development as evidenced by the wide fluctuation in feelings that is a hallmark characteristic of adolescence (Kuhn, 2009). Emotions serve as a vital function for motivation, behavior, and relationships (Kuhn, 2009). Learning to distinguish between functional and misleading thoughts and feelings is critically important in mental health. Emotional development needs to be understood in relation to the cultural and environmental contexts in which they occur, because specific social interactions influence learning (Zeidner, Matthews, Robers & MacCann, 2003). Illegal drug use is prevalent amount teenagers as a way to experience pleasure, reduce stress, and gain the approval of peers, among other reasons (Feldman, 2011).

Psychological changes. Adolescent psychological growth is characterized by identity development as youth seek to understand who they are and where they belong in the world (Feldman, 2011). Individuals start to look at themselves from a psychological or idealistic perspective rather than a physical one to make distinctions about who they are (Feldman, 2011). Knowing who you are (self-concept) and liking who are you (self-esteem) are two different concepts and lead to different outcomes. According to Erik Erikson (1968), adolescents discover themselves by experimenting with different roles and choices to see what fits for personal, occupational, sexual, and political commitments in a stage called identity versus identity confusion. Social comparison is another strategy utilized to clarify one's own identity. Those who discover their unique strengths and knowledge of themselves are better equipped to form relationships in the

next stage of development, intimacy versus isolation (Feldman, 2011). Negative outcomes include the inability to identify important roles or possible career paths (Erikson, 1968). More recent research indicates that identity development primarily occurs during the late teens and early to mid-20s, or during emerging adulthood, which is described in depth in the next section (Arnett, 2000).

Psychological problems, such as major depression and suicide, may occur alongside physical, cognitive, and emotional changes during adolescence. Of individuals aged 12 to 17, 2.2 million or 9% had experienced at least one major depressive episode with severe impairment (Substance Abuse and Mental Health Services Administration [SAMSA], 2014). The incidence rate is higher among females (19.4%) than among males (6.4%). The rate of suicide and self-injury is prevalent among adolescents and has increased in the United States in recent years (Muehlenkamp, Claes, Havertape, & Plener, 2012). Suicide is currently the second leading cause of death among 10- to 34-year olds, most prevalent among 18- to 25-year olds (SAMSA, 2014). The rates of suicide attempts are higher among females, but boys complete suicide more frequently (SAMSA, 2014). Those who are depressed, experience social isolation, perfectionism, high stress, and anxiety are at greater risk for committing suicide (Zalsman, Levy & Shoval, 2008). A recent study comparing suicidal thoughts, plans and attempts between adolescents and young adults with and without chronic illness found that suicidal thoughts and behaviors are higher among those with chronic disease, especially those with comorbid mood disorders (Ferro et al., 2017).

Social changes. The environmental and biological changes that occur during adolescence lead to new social situations and heightened awareness of and interest in other people. How well one manages emotions significantly impacts communication and is a key ingredient to developing rewarding interpersonal relationships (Choudhury, Blakemore, & Charman, 2006). Independence, or autonomy, and sense of control over life becoming increasingly important (Feldman, 2011). Adolescents transition from relying on parents for social support to their peers, making a sense of belonging important (Feldman, 2011). Conflict with parents is more common during the beginning of the teen years as one attempts to create emotional autonomy (Collins & Steinberg, 2006). Over time, the power dynamic becomes more symmetrical in the parent-child relationship, and thus less volatile (Whiteman, McHale, & Crouter, 2011).

Peer relationships are a hallmark characteristic of adolescence, and communicating with friends has never been more incessant or compulsive as it is today with the advancements in social media applications. Social comparison provides opportunities to evaluate one's own opinions, abilities, and attractiveness (Feldman, 2011). Peer groups serve as a reference for various roles and behaviors for the adolescent to try out in search of identity development (Feldman, 2011). Support from friends during adolescence is a protective factor and is predictive of both immediate and later resilient functioning (van Harmelen et al., 2017). The importance of belonging also increases the susceptibility to peer pressure, the process of conforming to peer attitudes and behaviors (Feldman, 2011).

Emerging adulthood development. The focus of the current study is on individuals diagnosed with JOMD during childhood or adolescence. To develop a comprehensive description of what people living with this chronic condition go through, participants were interviewed while in their 20s. As such, it is important to review current literature on this development period.

Jeffery Arnett's theory of emerging adulthood (2000, 2015) proposed that there is a developmentally and demographically distinct period between adolescence and young adulthood. The period of emerging adulthood takes place between the time of adolescence completion (usually around 18 years old) and the onset of stable roles in both love and work (Arnett, 2015). The age of completion varies between 25 and 29 years of age (Arnett, 2015).

Fifty years ago, most 21-year olds in America were married or engaged, done or almost done with an education, settled into a career or homemaking role, and ready to be parents. As such, the term "Young Adulthood" was more fitting, and was described by Erik Erickson (1950, 1968) as a life stage grounded in identity establishment, perpetuating the ability to make long-term commitments in intimate, reciprocal, relationships. The responsibilities of "adulthood" were clearly underway and within reach. Today, the average 21-year-old is more likely to be a college student, dependent on parental guardian(s) for residential or financial security, and about 6 years shy of marriage and parenthood (Arnett, 2015). This cultural shift occurred primarily in industrialized countries and opened a space for a new life stage: Emerging adulthood (Arnett, 2000, 2015).

Arnett (2004) described five distinct features that define emerging adulthood from other life periods. First, and perhaps most notably, it is "the age of identity exploration" (Arnett, 2004, p. 8). Though Erik Erikson (1968) described adolescence as the stage of identify versus role confusion, few 18-year olds today have entirely resolved the identity crisis. Identity exploration and formation intensifies and deepens during emerging adulthood. Young people work to establish a sense of who they are now, *and* determine what will bring satisfaction in the future, mainly through romantic and occupational pursuits with eventual long-term commitments (Arnett, 2004). Along the way, many individuals encounter perceived failures and disappointments, such as a relationship being terminated or being unsatisfied in one's selected college major. These experiences, as painful as they may be, can lead to a greater sense of self-understanding.

The second distinguishing feature of emerging adulthood is instability, or the high rate of change particularly in the areas of love, work, and residence (Arnett, 2004). Emerging adults are continually revising the "Plan with a capital P" (Arnett, 2004, p. 10), or path to successful adulthood (Arnett, 2004). The years between 18 and 25 are filled with more consistent modifications in romantic relationships, career decisions, and living arrangements than any other time in life (Arnett, 2004).

The third feature is the lack of obligation to others, leading to higher self-focus (Arnett, 2015). In other words, in efforts to become self-sufficient, emerging adults must learn how to live on their own without the guidance of parents or teachers. Therefore, daily life becomes more self-focused. Emerging adults are making their own decisions about tasks big and small—from what time to get up in the morning, to decisions about

attending college, or breaking up with a romantic partner. Becoming self-focused helps emerging adults gain a better sense of who they are and what they want out of life (Arnett, 2004).

The fourth distinguishing feature of emerging adulthood is the feeling of being "in-between" adolescence and adulthood. When asked, "Do you feel like an adult?" Arnett (2000) found that 60% of Americans between the ages of 18 and 25 answered "yes and no." The gradual transition can partially be explained by the three criteria that mark adulthood: accepting responsibility for oneself, making independent decisions, and becoming financially autonomous (Arnett, 2000; Greene, Wheatley & Aldava, 1992). By age 35, 70% of emerging adults believe they have reached "adulthood" (Arnett, 2004).

Emerging adulthood is "the age of possibilities" (Arnett, 2004, p. 16), or the time in life when an individual experiences a wide range of prospects for the future, and maintains high hope that his or her dreams will become a reality. Individuals have more freedom as a legal adult to make choices that reflect personal rather than familial values. In a *Time* National Study, 96% of 18 to 24-year olds agreed with the statement, "I am very sure someday I will get to where I want to be in life" (Hornblower, 1997). There is a lifetime high of hope towards one's future and ability to transform life (Arnett, 2015). Simply put, emerging adulthood is both a stressful and productive time of life, riddled with anticipation and execution of developmental tasks that have lifelong implications.

Emerging adults navigate five milestones, according to research, namely completing school/education, beginning a career, establishing independence, marrying, and parenting (Arnett, 2015, Setterstein & Ray, 2010). These transitions, in addition to

puberty-related hormonal changes, increased capacity for self-reflection, and changing familial and social relationships, may explain the rise in mental and emotional disorders (Ge et al., 2001, Koenig & Gladstone, 1998). During emerging adulthood, the presence of depression increased from 2% in childhood to 20% in adulthood (Costello, Erkanli, Fairbank, & Angold, 2002). In a nationwide study of 5.6 million Danish residents, Pedersen et al. (2014) found most substance abuse, schizophrenic and mood disorders, had peak ages of onset between the second and third decades of life.

Other studies have indicated that up to 25% of all emerging adults will experience a depressive episode by age 24, the highest incident rate of any adult age group (Jonas et al., 2003; Kessler & Walters, 1998; Klerman & Weissman, 1989). Anxiety disorders are also frequently diagnosed during emerging adulthood. Studies have shown a 5 to 19% occurrence rate (Pine & Klein, 2008), often with a co-morbid depressive (Sahoo & Khess, 2010) or psychoactive substance use disorder (Kessler et al., 1997; Morissette, Tull, Gulliver, Kamholz, & Zimering, 2007). These numbers are not expected to decrease anytime soon, as the pathway to adulthood is longer and more challenging than ever before (Setterstein & Ray, 2010).

Psychosocial Aspects of Chronic Illness and Disability

JOMD is one among many diverse chronic conditions that affect individuals in adolescence and emerging adulthood. What is common among them all is that disease onset elicits some type of response to the illness, and that one must adapt to the condition while continuing on the developmental trajectory.

Theoretical models of adjustment. Adjustment to disability research began in the 1950s and was originally conceptualized within a "coping" versus "succumbing" framework (Dembo, Leviton, & Wright, 1956). Essentially, researchers theorized that successful coping involved focusing on strengths as opposed to dwelling on the loss of function. Through personal accomplishments, control over one's life, and satisfying social activities, persons with acquired disability were considered to have a positive adjustment. On the other hand, poor adjustment is described as a continual focus on loss and the past while succumbing to a role as a helpless, incapable person (Dembo et al., 1956).

Over the last 60 years, researchers have developed many more models of adjustment, or having a good quality of life, well-being, vitality, positive mood, life-satisfaction, and self-esteem (Falvo, 2009). Livneh and Antonak (1997) defined adjustment as "a particular phase (i.e., set of experiences and reactions) of the psychological adaptation process" that marks the final phase of a crisis. Maladjustment, on the other hand, is characterized by perpetual anger, hostility, prolonged mourning, unnecessary dependency, and participation in self-destructive activities (Falvo, 2009). Those with a poor response feel ashamed of the illness, feel helpless, and have a negative body-image (Smart, 2009). Some experts do not agree with the term "adjustment" to disability because it implies something is wrong with having a disability, or that it means going through a series of stages to accept the situations (Toriello, Bishop & Rumrill, 2012). They prefer the term "adaption." For the purposes of this research review, adjustment and adaption are considered synonymous.

Understanding the process by which people adapt to chronic illness and disability helps inform the practice and treatment of counseling (Toriello et al., 2012). This section describes five theoretical approaches to psychosocial adaptation: The Stage Models, the Transactional Model of Stress and Coping, Ecological Models, The Disability Centrality Model, and the Peer-Grief Dynamic model.

Stage models. The stage models of adaption to disability/chronic illness conceptualize the response process occurring across four to six separate stages (Smart, 2009). Each is discussed in turn. The first stage is initial impact, or the initial reaction to a sudden disability (e.g., spinal cord injury) or the diagnosis of a chronic disease. Shock and anxiety are common emotions that have been described as overwhelming and can trigger panic (Marini, 2012a). Stage two, defense mobilization, is characterized by bargaining and denial, prevents "emotional flooding" as the individual gradually learns to assimilate the disability into everyday life (Smart, 2009). The third stage is grappling with the anger and depression that arise as the individual realizes the permanence and loss because of the disability (Marini, 2012a). Fourth is the retaliation stage, or a time when anger is externalized in the form of hostility to others, especially medical professionals (Livneh & Antonak, 1997). Non-compliance with treatment and blaming others and God delineate this stage. Finally, in the fifth stage—final adjustment or reintegration (also called integration or acceptance)—individuals are less distressed, as a new self-concept is formed (Smart, 2009). At this stage, the adaptation turns to actively mastering the environment and learning to effectively solve problems (Feldman, 2011). While the stage model offers insight into the adjustment process, critics argue that it is

impossible to predict everyone will go through the same stages, discounting coping skills, environmental factors, and other extenuating factors that play a role in adjustment (Marini, 2012b). A critique of the stage models is that they do not consider social barriers the disabled person faces and pathologizes inability to 'accept' the limited life caused by the impairment (Sapey, 2004).

Transactional model of psychosocial adaptation. The most well-known, frequently cited, and empirically supported theory of psychosocial adaptation is the Transactional Model developed by Lazarus and Folkman (1984). In this model, stress is the byproduct of the ongoing, constantly changing interactions between a person and the environment. Through primary appraisal, a person first determines the level of threat in each situation. If the stressor is congruent with goals/values, or considered to be beneficial, no coping mechanisms are needed and homeostasis returns (Marini, 2012b). If a threat is perceived (psychological, not physical), the individual moves to a secondary appraisal, where the options for coping are assessed. Questions such as "who is to blame?," "do I have any control to change the circumstance?" and "what could happen?" come to the forefront (Lazarus & Folkman, 1984). Two types of strategies are described: "Emotion focused" versus "problem focused" coping. Emotion focused coping refers to actions people use to soothe distress, such as social support, relaxation, or mindfulness, accepting that nothing can be done to improve the outcome. In contrast, problem focused coping utilizes strategies to troubleshoot the problem that created the stressor (Marini, 2012a). Eight groups of coping strategies have been outlined: confrontative, social support, distancing, self-control, escape/avoidance, reappraisal, problem solving, and

responsibility (Folkman & Lazarus, 1988). Variables such as personality traits, past experiences, and disease/treatment, serve as mediating variables in coping (Lubkin & Larsen, 2013).

Ecological models. The ecological models of psychosocial adaptation to disability highlight the significance of context in adaptation. The foundation for these models is comprised of three major tenets: (a) the nature of the disability, (b) personal characteristics, and (c) environmental influences (Marini, 2012b). Within each category, certain facets are described that have an impact on psychosocial adjustment. For example, in the first factor (i.e., nature of the disability), the sub-factors include the various nuances of chronic disease, such as time of onset, whether or not the disability happened suddenly or had a prolonged or gradual onset, functions impaired, severity of disability, visibility of impairment(s), stability of disease, and degree of perceived physical pain (Marini, 2012a). Personal characteristics, the second pillar of ecological models, are individual traits that influence adjustment. Factors such as gender, the activities impacted by the disease, values/goals, spiritual beliefs, and personal strengths/resources all contribute to the adjustment process and require assessment by a trained professional (Marini, 2012b). The third and final core piece of the model is examination of environmental influences on adjustment. Family acceptance and support is a significant determinant in positive adjustment (Waldboth et al., 2016). Other key sub-factors, including income, community resources, social support, and nursing home care, impact how well someone responds to disease and disability (Marini, 2012b). These models, most notably developed by Trieschmann (1988) and Vash and Crewe

(2004), have growing empirical support (Marini, 2012b). However, it is the Disability Centrality Model that has emerged in recent literature as the dominant model of psychosocial adaptation, because of its emphasis on perceived control of various domains that ultimately reflect life satisfaction (Bishop, 2005).

Disability centrality model. With its roots in Devins's (1994) concept of illness intrusiveness, and Dembo et al.'s (1956) emphasis on values, Bishop (2005) developed the most recent model of adaptation to chronic illness, the Disability Centrality Model, a quality of life-based framework of adaptation. This theory posits that perceived satisfaction in life is dependent upon how, and to what degree, a chronic illness or disability impacts domains that are important (central) to an affected individual (Bishop, 2005). While there is not agreement between what domains should be included, there has been increased agreement to include physical health, mental health, social support, employment or satisfying activity, and economic or material well-being as measures of quality of life (Bishop, 2005). The degree of control over one's medical treatment and environmental cconditionss is a significant aspect of the Disability Centrality Model. Increasing the sense of being in control and ability to master outside circumstances is central to establishing self-esteem (Zhou & Chen, 2017). Six domains encapsulate the theory: (a) Quality of life is subjective and multidimensional, representing an individual's holistic evaluation of important life domains; (b) those areas which are more important or highly valued to the individual will have more influence on perceived well-being compared to those of lesser importance (Bishop, Shepard, & Stenhoff, 2007); (c) people seek and desire to achieve and maintain optimal health, and the onset of a chronic illness

or disability initially reduces quality of life and feelings of personal control; (d) the number of life domains affected impacts the degree to which quality of life is reduced; (e) people naturally seek to minimize gaps between what is valued and what is realistic and (f) either strategies will be employed to change values and interests to increase life satisfaction, or nothing is done to improve control or force change (Bishop, 2005; Marini, 2012b). The Disability Centrality Model has been useful in improving the understanding of the impact of chronic illness and highlight the importance of self-management and perceived control among the multiple sclerosis population (Bishop et al., 2007).

While each of these models offers insight into the psychosocial adjustment to chronic illness and disability, they were developed by studying either congenital disabilities or one acquired later in life. JOMD is difficult to classify into either category —it is not present at birth nor is it an abrupt change, as in a spinal cord injury or amputation. There is already controversy as to whether person with disabilities eventually reach a stage of final adjustment or transcendence (Marini, 2012b). Living with JOMD is an ever-evolving process of progressive loss, hypothetically making adaption more challenging because the extent of disability grows over time. If there was such a thing as a final stage of adaptation, it is unclear if those with progressive neuromuscular disorders experience the eventual stabilization described in the literature.

Another limitation of the existing theories is that they were constructed with an adult sample. There is a dearth of literature regarding the experiences of adolescents and young adults with chronic illness, especially JOMD. Thannhauser (2009) was the first to

break ground and develop The Peer-Grief Dynamic model as a framework for describing the experiences of adolescents with multiple sclerosis (MS).

Grief-peer dynamics theory. Utilizing a grounded methodology approach, Thannhauser (2009) interviewed six 14–21 year old people diagnosed with multiple sclerosis and their mothers in order to provide a detailed description of the experience. The findings from this research study suggest that psychosocial development occurs within a context of grief. One moves through a cyclical process of grief and acceptance with peer relationships acting as a either a helpful or harmful force. Losses described were health, friendship, hope for the future, identity, control, and normalcy (Thannhauser, 2014). This is the first and only study that emphasized the role of grief as a key concept in pediatric chronic illness.

It is unclear if those living with JOMD would have a similar experience to those with MS. While both are incurable, chronic, and at times unpredictable, the nature, course, and compromised bodily system are different (i.e., MS degrades the myelin sheath on the nervous system; JOMD progressively weakens skeletal muscle). Since grief was a key concept in previous literature among adolescents with multiple sclerosis, current theories on loss needed to be explored in further depth in this study.

Theories on loss. The psychosocial adjustment models described in the literature review do not address the notion of loss. Two theories emerged in the current literature relevant to disability research: The Dual-Process Model (Stroebe & Schut, 1999) and the Meaning-Reconstruction Model (Neimeyer, 2000). Loss is an important concept in grief,

and is a risk factor for developing a mental disorder (Arroll & Howard, 2013; Prigerson et al., 1997).

Dual-Process Model. The Dual-Process Model was originally developed to describe a person's response to the death of a loved one (Stroebe & Schut, 1999), but is applicable to other types of bereavement, such as loss after a natural disaster (McManus, Walter, & Claridge, 2018) and loss of functioning associated with chronic illness (Miyabayashi & Yasuda, 2007). This model suggests that grief is a process whereby an individual oscillates between two orientations, one focused on the loss and the other on restoring or reorienting to a different circumstance (Stroebe & Schut, 1999). The loss-orientation is what most clinicians know as traditional grief work, or processing the emotional upset and longing that follows death or trauma (Stroebe & Schut, 1999). One then shifts into the restoration-orientation as a means of coping with or improving current circumstances, such as learning to pay bills if the deceased loved one managed the finances (Stroebe & Schut, 1999). A wide set of emotional reactions is involved in restorative tasks, from pride in mastering something new, to anxiety and fear of failure in going out alone.

The juxtaposition of loss and restoration captures the cognitive process that serves as a regulatory mechanism in confronting grief and avoiding it as a way of coping (Stroebe & Schut, 1999). Loss triggers unavoidable and sometimes conflicting responses, where a person must face or attend to aspects of grief, and then times when one can choose to avoid it, either by voluntarily suppressing emotions or involuntarily repressing painful feelings. Over time and repeated exposure to a certain aspect of loss,

habituation takes place and emotional reactivity is stabilized (Stroebe & Schut, 1999). The Dual-Process model provides a framework for understanding chronic grief. Pathology occurs when there is a disturbance in the oscillation process, resulting in stagnation in either grief or avoidance (Stroebe & Schut, 1999). The Dual Process model is an improvement from the stage model (described above) that posits that people pass through a series of phases that ends with acceptance, a construct that has been criticized as inappropriate for persons with chronic illness.

Meaning-Reconstruction Model. Neimeyer (2000) developed the Meaning-Reconstruction Model, which posits that significant events, such as loss, are opportunities for growth through re-learning what was once taken for granted. People have a natural inclination to develop meaningful self-narratives, which contribute to personal significance. In an ideal circumstance, one's internal perceptions of meaning are validated by one's social and cultural context (Neimeyer, Prigerson, & Davies, 2002). When a loss unexpectedly occurs, assumptions are threatened or destroyed and thus require rebuilding (Neimeyer, 2000). Newly constructed meaning is created over time and is unique to each person.

The Meaning-Reconstruction Model has its roots in constructivist assumptions and applies concepts from grief therapy. For example, one constructivist assumption is narrative truth, or that there is no one "true" reality that exists; rather, reality is individually constructed based on the storytelling used to make sense of life. Loss can either validate or invalidate assumptions we have about the experience. Another concept in this model, evolutionary epistemology, describes how the self-narrative changes over

time and with experience. Bereaved persons experiment with new ways of being until eventually finding a comfortable identity (Lister, Pushkar, & Connolly, 2008). Meaning reconstruction offers a contrasting view to the more commonly held belief that living with a disability is a tragedy.

To reconstruct meaning, one must complete three steps according to the Meaning-Reconstruction Model. First, people have to strive to find reasons "why" the loss happened. They are thrown into doubt and turmoil about what brings meaning and purpose to life at this sense-making stage, especially among those with chronic illness (Gillies & Neimeyer, 2006; Sim, Lee, Kim, & Kim, 2015; Zeligman, Varney, Grad, & Huffstead, 2018). The second step is benefit-finding, or building new narratives that incorporate—or are even build upon—the new circumstances created by the loss itself (Gillies & Neimeyer, 2006). The final step is identity change, whereby, in reconstructing meaning in loss, individuals reconstruct themselves (Gillies & Neimeyer, 2006).

Summary

Emerging adulthood is already a risk factor for the development of mental and emotional disorders, and there is compelling evidence to suggest that individuals with chronic illness are in greater danger of maladjustment. Many of the studies that have examined the experiences of emerging adults with chronic illness are with diseases that are more stable and treatable (e.g., arthritis can be treated with medicine). Much remains unknown about how emerging adults with JOMD experience the disease during this transitional and defining developmental period and navigate its course.

CHAPTER II

METHODOLOGY

The following chapter provides an in-depth description of the methods used in this study. First, descriptions of the researcher, sampling procedures, inclusion requirements, and participant selection are provided. Next, procedures and instruments for data collection are delineated, followed by a step-by-step description of the analysis. Finally, the trustworthiness considerations for this study are outlined.

A main tenet of qualitative research is that meaning is socially constructed by an individual's experiences and interpretations of the world around them (Merriam, 2002). One's "reality" cannot be objectively measured; rather, it is constantly in flux and changes over time (Merriam, 2002). Thus, qualitative researchers seek to understand another person's frame of reference, experiences, and meaning of an event (Merriam, 2002). This study, which investigates the experiences of emerging adults living with JOMD, was best conducted through qualitative inquiry. Furthermore, the openness of qualitative research is helpful when a phenomenon is poorly understood (Patton, 2002). The researcher could expand her understanding of the topic by asking the participant to clarify a response, checking the accuracy of her interpretations, or exploring unique reactions (Merriam, 2002). These two advantages (understanding the meanings attached to the phenomenon, and utilizing inductive methods to build understanding) led the researcher to undertake a qualitative design.

A descriptive phenomenological method best addresses the research question in this study, namely: How do emerging adults perceive and describe their experiences of

living with JOMD? Phenomenology is both a philosophy and a research method. The purpose of phenomenology in research is to understand and articulate a phenomenon, or lived experience, as it is experienced by a group of people (Giorgi, 2009). It is to understand what a specific phenomenon means from the purview of the persons who have had the experience and are able to describe it (Moustakas, 1994). From individual narratives, general themes or meanings are derived (Merriam, 2002; Moustakas, 1994). The researcher suspends all prior assumptions about the topic to understand it from the subjective world of the participant; the phenomenon is examined as it exists in the consciousness of the subject, not as it is in itself—or how it might be previously defined (Giorgi, 2009).

The philosophy of phenomenology is grounded in the notion that "Natural knowledge begins with experience and remains within experience" (Husserl, 1931, p. 9). The focus is neither on the human subject, nor the objective world; rather, a phenomenological approach seeks to understand how the individual interacts with and makes meaning of experiences (Merriam, 2002). This is the only true "reality" that exists (Merriam, 2002). In other words, to understand the experiences of emerging adults with JOMD, one must uncover the perspectives of those who have gone through the event. Semi-structured interviews assisted the participants in sharing their story. The open-ended nature of phenomenological research allows any and all dimensions of the experience to be brought to light. Aspect of living with JOMD, such as the negative emotions that emerge as the disease progresses, may have been lost in a quantitative design on pre-constructed instruments.

Bracketing Beliefs and Description of the Researcher

In qualitative studies, the researcher is the main source of collecting data, and thus plays an essential role in—and is influential to—the investigation process (Converse, 2012). A skilled researcher is one who is knowledgeable about the research topic, adept at listening, observing, responding, and adapting to the evolving narratives of participants to gather rich, descriptive data that is verbal as well as non-verbal in nature (Merriam, 2002). Experience with human interactions and proficient communication skills are paramount to accurate and genuine responses (Bellini & Rumrill, 2009). The researcher had a keen understanding of the topic under investigation because she lives with JOMD. This personal experience helped in understanding the complexity of the condition. While the question of scientific inquiry emerged from personal experience, it has social meaning for others affected by JOMD (Moustakas, 1994).

While personal history inspired the research question, it also created biases that could slant the interviews and analysis (Moustakas, 1994). The researcher engaged in the epoché process prior to data collection and analysis (Giorgi, 1997). Epoché describes the state where all preconceived notions about a topic are suspended to approach, observe, and understand a phenomenon as if one has never seen or experienced it before (Giorgi, 1997). One refrains from perceiving it in an "ordinary" way; rather, the phenomenon is studied from a fresh, naïve vantage point each time (Moustakas, 1994). To accomplish this, the researcher responded to all research questions and reflected on her experience herself to highlight possible assumptions and expectation. These past experiences were then "bracketed," or "put aside" before each interview to allow the unique experience of

living with JOMD be explained by each participant without any impositions from the interviewer (Merriam, 2002). Giorgi (2009) described bracketing as "holding in suspension" (p. 93) the past and the present, or attempting to notice one's biases when making judgments about the data. Bracketing helped present personal biases and reduce the potential to gravitate toward similar data (Hatch, 2002).

Researcher

The researcher for this study is a mid-30s Caucasian female, who resides in a suburb of Northeast Ohio with her husband and pet dog. She and one of her four siblings live with LGMD2a. She was diagnosed with LGMD2a at the age of 18, her brother at age 13. No other extended family members are affected with any form of MD to her knowledge. Over the course of this study, the researcher transitioned from no assistive technology to utilizing a power wheelchair for more than eight hours a day due to LGMD progression.

The researcher is a doctoral candidate in the Education, Health and Human Services program at Kent State University in Kent, Ohio. She is an independently licensed professional clinical counselor with a supervision endorsement, working full-time, in a private practice setting counseling individuals aged 12 and up. The researcher also has experience at a university counseling center, and in a hospital's intensive outpatient program and adult psychiatric and psychological treatment services. She has experience teaching graduate and undergraduate courses at local universities.

JOMD Experience

The researcher has lived with LGMD2a for 17 years, and it has been a part of her family for 25 years. She was diagnosed five years after the first noticeable symptoms of muscle weakness. Emerging adulthood was a tumultuous time from an emotional and physical standpoint as she realized her dreams of being a nurse were incompatible with declining muscle strength. With the help of mental health counseling, she successfully reintegrated back into higher education and found a meaningful and suitable career. Processing her grief of living with JOMD in therapy was life changing. Since that time, she has volunteered with the Muscular Dystrophy Association (MDA) for four years and participated in clinical trials for LGMD. She currently meets with local families who are affected by JOMD to provide education about the diagnosis and help in the adjustment process.

Hence, her experience of living with LGMD2a sparked an interest in examining the impact JOMD had on other emerging adults to understand how counselors may be more active with and helpful to this population. Assumptions of the topic include the following:

1. There are common experiences shared by people who obtained a diagnosis of JOMD.
2. Individuals with JOMD experience emotional distress during emerging adulthood, especially as the disease progresses.
3. As the disease progresses, it is stressful and the individual has to continuously adapt.

4. Medical professionals do not assess emotional health.
5. Individuals with JOMD experience a disruption in one or more developmental tasks.
6. The disease impacts the family, especially the parent(s).
7. Individuals with JOMD will report seeing life differently because of the illness, both in positive and negative ways.
8. The affected individual has periods of perceived isolation because of the disease.
9. Mental health counseling will be a helpful intervention, particularly during emerging adulthood.

Sampling Procedure and Participant Selection

Determining sample size in qualitative research is a nebulous concept (O'Reilly & Parker, 2012). There are no rules of thumb to determine a precise number of participants (Patton, 2002). Rather, it depends on the qualitative concept of thematic or data saturation, or the point at which no new data or themes are being generated (O'Reilly & Parker, 2012). The purpose of this study was to illuminate how emerging adults perceived and described their experiences of living with JOMD. Thus, the researcher focused on understanding all that is important about that experience from those who have lived it (i.e., a broad range of experiences for a small number of people). It is up to the researcher to determine how long to remain in the field, a decision reached after examining the data, themes, and interpretations time after time to ensure they make sense and represent the participants' realities (O'Reilly & Parker, 2012). At the onset of the

study the sample size was estimated to be between 4 and 12 participants. Giorgi (2009) recommended at least 3 participants to obtain divergent perspectives, multiple frames of reference, and to be able to fully describe the experience (i.e., adequate imaginative variation). A total of eight individuals were interviewed.

Purposeful sampling was utilized to recruit research participants. Because the researcher sought to gain an in-depth understanding of a rare medical condition, this sampling method was the most effective (Patton, 2002). Criterion sampling (a form of purposeful sampling) was most appropriate for the study. Criterion sampling involves the researcher finding individuals who meet certain criteria (Given, 2008), which is discussed shortly.

Gaining access to participants proved to be difficult. Initially, one key informant from a local Muscular Dystrophy Association (MDA) office, Jackie Becker, dispersed information about the study via an MDA sourced listserv. This recruitment route provided a total of six people, only three of which met the recruitment criteria and were interviewed. After two additional "re-posts" of the study, the researchers (and the key informant) ceased advertisement. New means for participant recruitment were needed.

Six months after the initial IRB approval, the researcher requested and was granted a second approval for an IRB amendment to improve the study advertisement and add more methods for recruitment. A combination of social media and addition of a key informant provided enough volunteers to reach data saturation. Facebook was chosen as the best social media platform, because it already offers several "groups" people may join if they have MD (to name a few: "Limb Girdle Muscular Dystrophy" and "Beyond

Labels & Limitations"), thus providing a platform for those affected to discuss MD related events. The study advertisement was posted on various MD Facebook Groups' pages. The additional key informant, Jodi Wolff, PhD, has been in the neuromuscular field for over two decades. She was willing to pass on the study advertisement to her wide network of personal friends and family members who potentially meet recruitment criteria (as opposed to sharing private contact information with the researchers). Five more individuals came forward to be interviewed at which point the researcher decided to stop the recruitment process.

Selecting individuals to interview for this study was based on the following criteria. A more detailed description follow.

1. Were between the ages of 18 and 29.
2. Had no known cognitive impairments or problems understanding English that would significantly impair his or her ability to participate in the interview.
3. Willing to participate in two 60-90-minute audio-taped interviews.
4. Had a form of JOMD, such as Limb Girdle, Distal, Becker's, Facioscapulohumeral or Emery-Dreifuss, or a type that was diagnosed in later childhood or adolescence.
5. Did not have 'Birth/Early Onset' forms, such as the Duchenne type.
6. Could recall memories of living both with and without the condition (or when the disease became noticeable to them).
7. Had been diagnosed with MD for at least one year prior to participation.

The first inclusion criterion was that only emerging adults—or those between the ages 18 and 29—were included. To capture the experience of living with JOMD in emerging adulthood, the researcher chose to interview only those in this life stage to discover the meaning of the experience in "real time" as opposed to retroactive reflections of varying degree.

The second and third inclusion criteria were added to ensure rich, detailed, and complete descriptions of the phenomenon under investigation (Merriam, 2002). Linguistic or cognitive barriers could interfere with accurate and complete data. Thus, if there were problems understanding or speaking English, the person was not included. Participants were not excluded from the study based on gender, religion, sexual orientation, physical ability, race, or ethnicity.

The participants agreed to participate in two 60-90 minute interviews. The time limit was established intentionally, to provide enough time for comprehensive descriptions, but not be too long to have the participant drift away from the specific experiences of living with JOMD in emerging adulthood, or to exhaust the participant.

Criteria 4, 5, and 6 are the exclusive criteria put in place to add an innovative perspective to the existing literature. This study examines a specific group of the MD population, one that was identified based on reviewing the literature on lifespan development, chronic illness, and mental health. Participants all had to have a form of JOMD, defined in this study as (a) being diagnosed with MD in late childhood or early adolescence; (b) not having the "Birth/Early Onset" forms such as the Duchenne type, and (c) having the ability to recall memories of living both with and without the

condition, to describe the progressive aspect of the illness, both physically and psychologically.

Finally, the participants needed to have been officially diagnosed with a type of JOMD (e.g., BMD, LGMD, EDMD, FSHMD) by a medical doctor no less than one year prior to participation. This restriction was put in place to (a) protect the well-being of participants, and (b) promote thoughtful description of the experience. The nature of this topic is highly sensitive and personal. Without at least one year to adjust and process the diagnosis, it could have been too painful to describe. This decision was grounded in research suggesting that the initial shock and grief typically subside within one year (Smart, 2009). Interviewing participants soon after diagnosis could have resulted in data focused more on shock and emotion regulation. Furthermore, to describe "living with" the disease, adequate time must have been provided for the participant to experience life after diagnosis. The 12-month criterion allow for greater reflection and clarity in describing the experience, two qualities necessary in phenomenological research (Merriam, 2002).

The sample for this study is comprised of eight people, of which three were recruited from the initial key informant, five from Facebook. Seven of the eight individuals are living with a form of LGMD, and one has EDMD.

All elected participants were first screened to ensure inclusion requirements, and gave their consent to participate in the study. Each participant agreed to be audiotaped during the interviews and was informed that the interview could be terminated at any time without consequence. To ensure confidentiality, pseudonyms were utilized and

attached to each interview transcript and subsequent analysis procedures. The interview transcripts, meaning units, and transformations of the text were kept in a secure location and password protected electronic files.

Procedure

The following section delineates the research process. Participant recruitment and data collection are described, followed by a presentation of the instruments utilized in this study, including screening and demographic forms, interview questions, and attainment of field notes.

Participant recruitment. The very first step in this−and any other ethically sound research study using human subjects—was to gain written approval from the Kent State University Institutional Review Board (IRB; see Appendix A). The IRB serves to protect the welfare and rights of all individuals who participate in research. This study was granted initial IRB approval on June 17, 2016. Another IRB Amendment was granted on December 22, 2016, to include modifications for participant recruitment procedures (i.e., adding an additional key informant and permission to post advertisement on Facebook) and edits of the original study advertisement; it can be viewed in Appendix B.

Following IRB approval(s), the researcher contacted key informants via email (Appendix C & D) to describe the purpose of the study and to request assistance dispersing the advertisement via listserv or to personal contacts known to potentially meet recruitment criteria (i.e., purposeful sampling). The researcher also posted the

advertisement on various Facebook "group" pages comprised of members with LGMD (Appendix E).

Upon first contact, the researcher gathered more detailed information from eligible participants (see Appendix F; Screening & Demographic Form). These documents were kept in a secure location (e.g., locked filing cabinet in 310 White Hall).

Data collection. Schram and Schram (2006) noted that one of the assumptions of a phenomenologist is that the meaning of an event (i.e. phenomenon) can be discovered through dialogue and reflection. Thus, the main source of data was interviews, or the participants' description of their experiences. This is the single best way to obtain the lived experiences that belong to each person (Giorgi, 1997).

Participants were given the option to be interviewed either face-to-face, by phone, or via Skype, an internet-based video communication system. The participant who chose an in-person interview completed the informed consent (Appendix G) and consent to audiotape (Appendix H) documents at the time of the first interview. If participants were not meeting with the researcher in person, they were mailed or emailed a copy of the informed consent document and consent to audiotape. Once these documents were completed and returned, the first interview was scheduled and recorded over the phone.

For the purposes of this study, the main source of data was collected through interviews. The researcher approached each interview with a tentative, "not-knowing" disposition known as epoché, or an intentional effort to be completely open and perceptive to listening to each participant's story as it unfolds (Moustakas, 1994).

Preconceived notions about the phenomenon under study were bracketed, or set aside, to be fully present during the interview (Giorgi, 1997).

Broad and open-ended questions were prepared to elicit thick, rich descriptions of the phenomenon (which provides the foundation for phenomenological analysis; Patton, 2002). However, Moustakas (1994) stated that

> Although the primary researcher may in advance develop a series of questions aimed at evoking a comprehensive account of the person's experience of the phenomenon, these are varied, altered, or not used at all when the (participant) shares the full story of his or her experience. (p. 114)

The inner experiences of participants emerged during this fundamental and crucial first step. All interviews were recorded and transcribed.

At the outset of the first interview, the researcher completed a Participant Data Sheet (Appendix I). Completing the Participant Data sheet with the participant provided an opportunity to establish rapport before the interview began. A conversational approach was taken to engage the interviewee in a partnership that supported and encouraged their story (Rubin & Rubin, 2005).

A minimal amount of self-disclosure on behalf of the researcher helped to minimize the power dynamic, and assist the interviewee in feeling protected and comfortable (Creswell & Miller, 2000; Rubin & Rubin, 2005). This helped to build empathy and establish rapport, two conditions which must be met for qualitative research (Giorgi, 2009). Due to the deeply personal and self-revealing experiences that were

articulated, the researcher spent extra time building rapport during the initial interview. Giorgi (2009) encourages these bonds before data collection in a study of this nature.

The interview proceeded with descriptions of living with JOMD. Guiding questions elicited descriptions of how and when JOMD first entered the consciousness of the participant, and how the experiences of living with JOMD evolved as the condition progressed. The interview questions are available in Appendix J, and described in more detail in the Instrumentation section.

Participants were encouraged to broadly describe their experiences via the initial interview question: "How would you describe your experiences of living with your condition, beginning with your first memories up until now?" Guiding questions helped the participants "tap into" the experience and promote comprehensive descriptions of the phenomenon (Moustakas, 1994, p. 116). The researcher then transcribed the interview. The audio-recordings were listened to several times, and transcripts were read repeatedly to get a general sense of what the interview had produced in terms of data. This step helped the researcher gain a sense of the entirety of the data, before it was deconstructed into meaning units and themes (which is described in detail later; Giorgi, 1997). Furthermore, transcribing and reviewing the data provided the researcher an opportunity to further reflect on the first interview and become aware of missing or unclear information before the second interview. Please see Figure 1 for the research process flowchart.

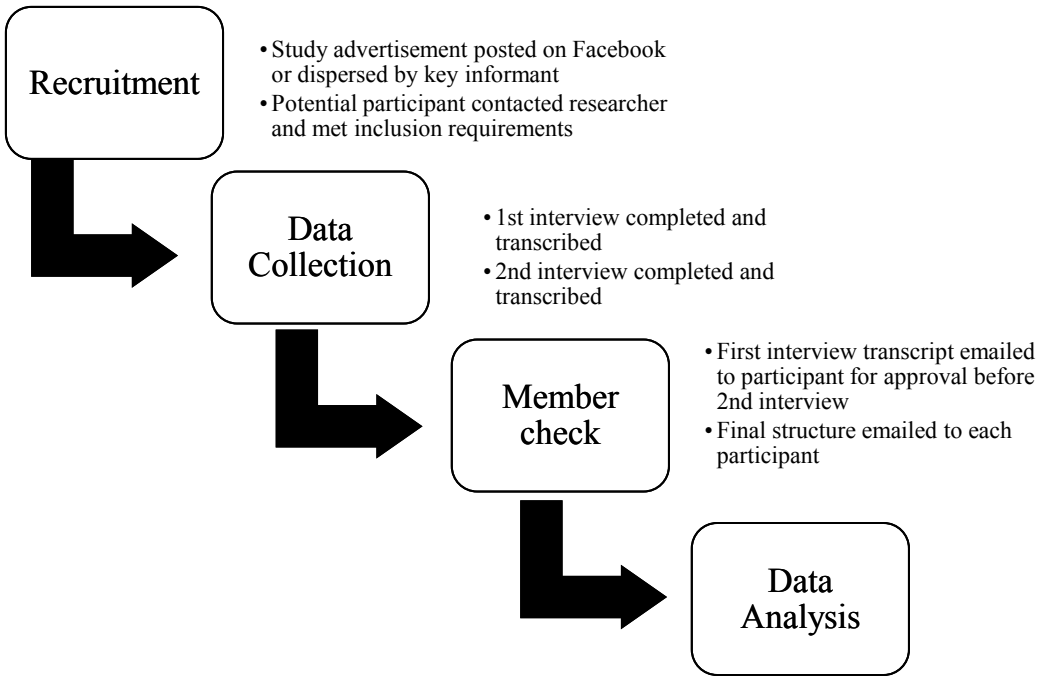

Figure 1. Research Process Flowchart

Each participant was emailed a copy of the first interview transcript to check for accuracy. This is known as a "member check" (Lincoln & Guba, 1985). The data must be confirmed by the original source for the study to be credible (Lincoln & Guba, 1985). Member checks are described in detail in the trustworthiness section. The second interview was scheduled after the participant reviewed the transcript and confirmed its accuracy. If any changes needed to be made, the participants were instructed to contact the researcher via phone or email. Once the transcript and themes were deemed accurate, the second interview was scheduled.

The purpose of the second interview was to fill in any information that was missed during the first interview, and expand and elaborate on the research topic (see Appendix K for Second Interview Questions). Participants were also encouraged to share

specific concepts that he or she wanted the researcher to emphasize. This highlighted what stood out to them the most, thus proving to be an important aspect of the phenomenon. A second interview also allowed the participants to respond to the researcher's interpretations and summary of the first interview (Lincoln & Guba, 1985). In other words, "It puts the respondent on record as having said certain things and having agreed to the correctness of the investigator's recording of them, thereby making it more difficult later for the respondent to claim misunderstanding or investigator error" (Lincoln & Guba, 1985, p. 314). Sharing these preemptive results is another form of participant checking (Lincoln & Guba, 1985).

Counselors uphold the ethical standards outlined in The American Counseling Association's (ACA) Code of Ethics (2014), one of which states, "Counselors act to avoid harming their . . . research participants and to minimize or to remedy unavoidable or unanticipated harm" (A.4.a., p. 4). To ensure this standard, time was set aside during the first and second interviews to process any difficult emotions that emerged because of the study. It was important that no participant was left in a state of distress. The researcher inquired about the experience of being interviewed, and how the participant was feeling as the interview concluded. The researcher provided referrals to local mental health agencies (see Appendix L) if the participant expressed interest in further processing their experiences or demonstrated symptoms of a mental health disorder. Each participant was contacted after the second interview to address any questions or concerns from the study.

The second interview was transcribed within three weeks. The researcher again listened to the audio-recordings several times and checked the transcript for accuracy. Data analysis resumed. New input was incorporated into the original data from the first interview to provide a detailed description of the phenomenon. A peer auditor was utilized to confirm results and ensure proper analytic proceedings. Each participant was asked during the final interview if he or she could be contacted via email (Appendix M) to respond to the final analysis. All participants confirmed the findings. See Appendix N for a detailed description of the research schedule.

Instrumentation

Three sources were utilized to gather information for the study: (1) demographic forms, including (a) the Screening Form and (b) the Participant Data Sheet; (2) first interview schedule, and (3) second interview schedule and follow up email. A detailed description of each is provided in this section.

Demographic forms. Two documents were completed by the researcher to gather information about participants: the Screening Form and the Participant Data Sheet. These data contributed to providing a detailed description of the sample subject. While qualitative research cannot be generalized, this material aids in the study's transferability to the audience (Lincoln & Guba, 1985).

Screening form. The screening form may be viewed in Appendix F. It served to record eligibility data, such as date of birth, type of MD, date of diagnosis, and physician who determined diagnosis. If the person met recruitment requirements and was willing to

participate in two 60–90-minute interviews, the researcher gathered contact information (e.g., address, email) and possible dates and times for the first interview.

Participant data sheet. The participant data sheet recorded further demographic information. Participants provided a description of race, nationality and/or ethnicity, gender, employment status, highest level of education, relationship status, who lived with subject at the time of diagnosis, who lives with the subject now. If the individual had any questions about the form or participating in the study, he or she was encouraged to ask.

First interview schedule. Verbatim interview transcripts from the first, semi-structured, interviews were the second instrument utilized for this study. Patton (2002) described such transcripts as the essential raw data for analysis. The purpose of the interview was to understand the lived experiences of each participant (Merriam, 2002). No other data set were more critically important to the outcome of the study than the first interview transcripts. The researcher developed seven central research questions to understand the topic under investigation. Each was designed to maximize participant responses. Sub-questions were altered, added, or subtracted, depending on participants' answers. If responses were confusing or incomplete, the researcher sought clarification by paraphrasing what had been said, or by asking a follow-up question. The researcher designed the following questions for the first interview to help the participant fully describe the experience of being diagnosed and living with JOMD:

1. How would you describe your experience of living with your condition, beginning with your first memories of the disease up until now?

   a. What aspects (being diagnosed, loss of ability, etc.), occurrences, or people connected to the experience stand out to you?
   b. How does having MD affect you and your life? How has it changed you?

2. How do you believe having your condition has affected significant others in your life?
3. Who, if anyone, do you lean on for support in coping with your condition?
4. What is the most difficult aspect of having this diagnosis as an emerging adult?
5. What, if anything, do you believe could be helpful to you to better cope with your condition?
6. How do expect your condition will impact your future?
7. How did you get more information about your diagnosis (from a parent, doctor, self)? When did you learn it? What was it like?
8. Have you shared all that is significant about your experience of living with this diagnosis?

Second interview schedule and follow-up email. The purpose of the second interview was to fill in any missing information, to discuss emergent themes from the first interview (i.e., conduct a member check), and if necessary, to clarify the meaning of a participant's response. The second interview afforded the researcher the opportunity to (a) explore the accuracy of themes for each participant, (b) confirm and authenticate

findings, and (c) gather supplementary information about the experience of living with JOMD in emerging adulthood.

1. Each participant was asked the following questions: What thoughts/feelings emerged for you as you reflected on our first interview?
2. What stood out for you the most from our first interview?

The researcher then shared a summary of the data from the first interview. The researcher frequently paused to allow the participant to contribute his or her thoughts. While the summaries were being read, the participant reflected on how well the summary matched up against their own thoughts and experiences. The final questions were as follows:

3. Has anything been excluded about your experiences of living with this diagnosis?
4. Have you shared all that is significant about your experiences of living with your condition?

Each participant agreed to read and respond to a follow up email containing comprehensive findings (Appendix M). The email shared themes that resulted from the accumulation of data from all participants. Each participant was asked to reflect on the findings and offer feedback.

Analysis

A procedure by Giorgi (1997, 2009) was employed for analysis of the verbatim transcripts. The five basic procedures as are follows: (a) read for a sense of the whole, (b) determination of meaning units, (c) transformation of meaning units from

participants' verbatim responses into phenomenological expression, (d) organizing and expressing the data into integrated narrative, and (e) synthesis or summary of the data to communicate to the intended audience. An elaborate description of each step is provided after Figure 2, visual description of the Data Analysis Process.

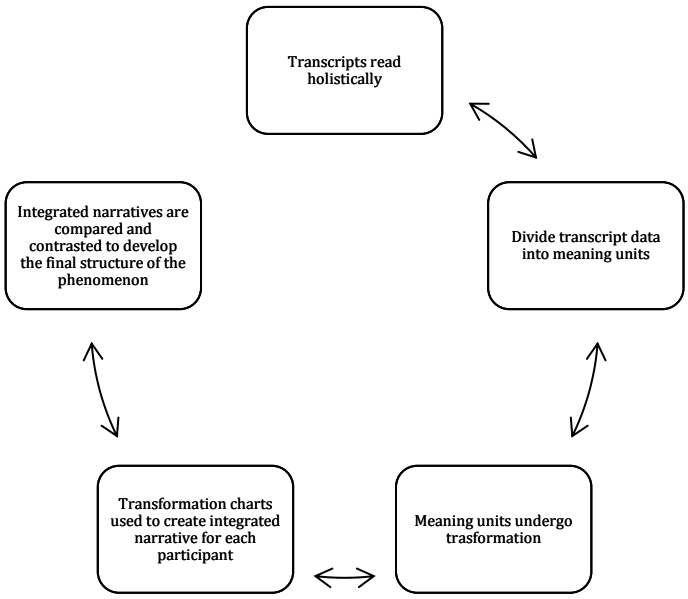

Figure 2. Data Analysis Process

First, all interview transcripts were read to acquire a global understanding of the material. Phenomenology is holistic—thus one must read all the data and gain a sense of how it fits together before it is broken down into meaning units and themes (Giorgi, 1997, 2009).

Second, the data were divided into parts, or meaning units, that address and describe the experience under investigation. To discover meaning units, the author approached the interview transcripts with an open, "discovery-oriented" disposition to "let unexpected meanings emerge" (Giorgi, 1997, p. 130). Each interview transcript was

re-read slowly from beginning to end, and the researcher documented each time there was a shift in meaning when the participant was describing life with JOMD (Giorgi, 2009). By the end of this step, the researcher had an arbitrary set of meaning units, or descriptions of the phenomenon, in the participants' own words (Giorgi, 1997, 2009).

Every expression that was not relevant to the experience of living with JOMD was eliminated. This process is also known as "reduction and elimination," whereby the researcher selects only phrases relevant to the experience of living with JOMD, and eliminated all phrases that did not describe the phenomenon (Moustakas, 1994, p. 121).

Third, the researcher transformed the raw data (i.e., participant's everyday language) into phenomenologically and psychologically pertinent expressions (Giorgi, 1997, 2009). The researcher went back to the meaning units and transformed each one from the participant's raw expression to a more satisfactory statement reflecting the psychological implications of lifeworld description (Giorgi, 2009). The first transformation began with restating each meaning unit by changing it from first person to a third person perspective and clarified, but keeping much of the original language intact. This ensures the data is from the participants' and not the researcher's experiences. The first transformation was done within the original transcript to ensure accuracy and correct context. The raw statements and first transformations were organized chronologically and coded (i.e., diagnostic experience, fear of the future). Each meaning unit underwent a third transformation, whereby the researcher carefully detected, drew out, and elaborated on the psychological experience of living with JOMD by rephrasing the direct quotes into more straightforward, clear statements (Giorgi, 2009, p. 131). The meaning

units are carefully transformed into psychologically pertinent expressions that eventually help to illuminate the structure of the phenomenon (Giorgi, 2009, p. 137). Free imaginative variation allowed the author to examine the data from different perspectives and create higher-level categories that retain psychological meaning and reveal characteristics of the phenomenon. See Table 2 for an example of the data transformation process.

Table 2

Data Transformation Example

Raw data	1st Transformation	2nd Transformation
"Also, because the symptoms of not being able to - or losing the ability to walk was also the . . . I think it was the key too for which caused the panic attacks. This I think it was really it hit a point in deep inside the unconscious brain or I don't know how to describe it better but I think it would hit the point which then created these panic attacks to alert myself too that I shouldn't be scared to take breaks regularly or to use the wheelchair or to talk to parents to change the house because there will be one day when my ability or the ability would have been gone, of walking so I really found out for myself that if I start to struggle with—to lose movement or I don't know to lose the capacity of my arms or whatever then there always should be one step ahead and already have a solution to count on. To not be in this kind of situation again to create fear."	She believes the progression of LGMD and losing the ability to walk also caused the panic attacks. This hit a point deep inside her unconscious brain that created panic attacks to alert herself that she shouldn't be scared to take breaks regularly, use the wheelchair and talk to her parents about changing the house because one day her ability to walk will be gone so she found out for herself that if she is struggling or progressing she should always be one step ahead and have a solution to count on so she does not have to be in the situation again to create fear	The fear and growing awareness that LGMD was progressing and that she was losing the ability to walk was a main contributor to the panic attacks. She was scared to use a wheelchair though her legs needed breaks, and she was scared to talk to her parents about changing the house to make it more accessible. She knew one day her ability to do these things would be gone, and she didn't know what the solution was.

Fourth, the researcher synthesized the 3rd transformations from each participant into an Integrated Narrative, describing the participant's experiences of living with JOMD. The Integrated Narrative formed a cohesive description of the phenomenon from each participant's perspective. It also generalized the data to a certain degree, so that it was easier to integrate the data from various participants into one structure (Giorgi, 2009). While facts of the experience of living with JOMD differed between participants, there were identical psychological meanings. Essential elements emerged, and were compared for intersubjective agreement. Themes were generated and described.

This leads to the final step in data analysis, which is expressing the structure of the phenomenon (Moustakas, 1994). The Integrative Narratives and transformed meaning units form the foundation for writing the structural description of the experience, which consists of key elements and the relationship between them (Giorgi, 2009). In the process of writing the structure, aspects of the phenomenon, which the participants may have been unaware of, are described. Implicit meanings across participants that were not directly stated but had a strong background presence were made explicit. In addition, the researcher attempted to make the data transferrable so that readers can integrate the information (Giorgi, 2009). For example, each theme was considered in both broad and specific terms. Participants' narratives helped to explain their point of view, but sweeping statements were made to make it applicable to others with JOMD.

It is important to note that data collection overlapped with analysis, so that the author could adjust guiding questions to maximize participant responses. Tentative

categories and relationships were formed; waiting until the end of the data collection to begin contemplating analysis, would result in a great deal of missed opportunities to fully understand the phenomenon. This also illuminated a stopping point. By constantly reviewing and analyzing data in transcripts, interviews ceased after the researcher found nothing new in the data, only repetitions of what was already known (Hatch, 2002).

Trustworthiness

For a phenomenological inquiry to be regarded as trustworthy, four conditions must be met. Lincoln and Guba (1985) described these prerequisites as (a) credibility (internal validity), (b) transferability (external ability), (c) dependability (reliability), and (d) confirmability (objectivity). Each are discussed in turn.

Credibility

Credibility in qualitative research means the study accurately presents the phenomenon under investigation (Shenton, 2004). In other words, and in this case: Does this study justly describe the experiences of emerging adults living with JOMD? To establish strong credibility for this study, several strategies were promoted. The first is triangulation, or using different sources to confirm findings (Merriam, 2002). Without verification from several sources, no report is credible (Lincoln & Guba, 1985). Multiple interviewees provided diverse and rich descriptions of the same phenomenon, which were all utilized in data analysis. By having verification for a single theme from several participants, the results suggest commonality and credibility.

Honesty on behalf of participants is a critical component of credibility (Shenton, 2004). The researcher established informed consent and encouraged genuine responses

followed with empathic listening. By providing participants with the option to refuse responses to any questions, or terminate the interview at any time, researcher ensured that only those who were genuinely willing to discuss the experiences participated in the research (Shenton, 2004). The researcher made it clear that the participant had the right to withdraw from the process at any point without consequence.

Another method to build credibility was member-checks, or verifying the researcher's data and conclusions with participants (Lincoln & Guba, 1985; Shenton, 2004). Member checks and sharing the final themes offered participants the opportunity to confirm that the data and results were an authentic explanation of the phenomenon. Lincoln and Guba (1985) described member-checks as "the most crucial technique for establishing credibility" (p. 314). Thus, the researcher requested member-checks at three points during the study: (a) participants verifying the accuracy of verbatim transcripts, (b) sharing a summary of the first interview data and confirming emergent themes during the second interview, and (c) a follow up email authenticating the findings of the study. A detailed description of the experiences of participants with MD was provided. It "rang true" for the participants (Merriam, 2002).

Lastly, a peer auditor was utilized was to ensure credibility by examining the audit trail materials, or raw data, data reduction, transformation tables, integrated narratives, reconstruction, and final synthesis. Lincoln and Guba (1985) recommended the auditor be a peer of the primary researcher; this is a person that is not in a position of authority (e.g., dissertation committee chairs), but skilled in the methodology through basic and advanced graduate level training in qualitative analysis (Lincoln & Guba,

1985). The peer auditor for this study was hand-selected because he was formerly a peer of the researcher but completed his Ph.D. in Counselor Education and Supervision in August 2014. Furthermore, he has experience with Giorgi's phenomenological methods. He is now an assistant professor in a Counselor Education program.

Transferability

Transferability refers to the extent to which results from one study can be applied to other contexts or situations (Merriam, 2002). In qualitative research, it is impossible to establish external validity (i.e., generalizability) in the conventional sense (Lincoln & Guba, 1985). The researcher can only provide sufficient contextual information so that the reader can make judgments as to the "fittingness" of the results to other conditions (Lincoln & Guba, 1985). In other words, by providing rich descriptions of the participants and their experiences, the reader himself can decide if the results apply. Thus, the author provided ample descriptions of each participant so that a decision of transferability can be made independently.

Dependability

Dependability, or reliability, suggests that if the study were repeated, similar results would be achieved (Shenton, 2004). Like transferability, this does not align with the purposes of qualitative research. The nature of the phenomenon may change or may be specific to the participants of this research. Nonetheless, the researcher provided an explicit depiction of research design, procedures and implementation; a future investigator will have the tools to replicate the inquiry undertaken in this study.

The researcher utilized an inquiry audit to establish dependability (Koch, 1993; Lincoln & Guba, 1985). Essentially, the peer auditor followed the decision trail of the primary investigator and arrived at comparable conclusions (Koch, 1993). The auditor examined both the process of how conclusions were drawn, and the products themselves (e.g., data, journal entries, findings, interpretations; Lincoln & Guba, 1985). It "verifies the bottom line" of the study (Lincoln & Guba, 1985, p. 318).

Confirmability

An inquiry audit cannot be conducted without the researcher first recording summaries, interpretations, and researching findings—otherwise known as an audit trail (Lincoln & Guba, 1985). This helps establish confirmability, which in qualitative research emphasizes the importance of objectivity (Shenton, 2004). In its purest form, a qualitative researcher cannot be completely objective; the intrusion of the researcher's bias is unavoidable (Shenton, 2004). Thus, the auditor helped to abate these natural flaws. The researcher also bracketed, or did her best to set aside her presumptions of the experience of MD, to approach inquiry with an open disposition (Giorgi, 1997). Triangulation is an important technique to emphasize again in establishing confirmability (Lincoln & Guba, 1985). Finally, confirmability will likely be established when credibility, transferability, and dependability are attained (Koch, 1993).

Peer Auditor

The results of this study are not only the work of the researcher, but also the peer auditor, a former student in the Counselor Education and Supervision program at Kent State University and current professor. The peer auditor in this study helped the

researcher develop the dissertation document by questioning methodological procedures that led to increased transparency and clarity in the analysis section. Correspondence occurred via email. More specifically, the peer auditor read the dissertation proposal and questioned how the researcher will transform the individual narratives into one integrated phenomenological essence. He also inquired about questions asked of participants to verify the data.

Regarding analysis, the peer auditor reviewed the raw data to assess the plausibility of themes, and found the findings to be true to the narratives of participants. He provided feedback theme by theme, communicating what stood out and where the researcher needed to add more depth and complexity. His feedback inspired the section on compounded losses and reticence of being in the role of a spokesperson for JOMD. He highlighted gaps in the themes that required further explanation or quotes to support it, such as in fear of the future. The peer reviewer also provided different ideas about the stressful nature of JOMD that would be relevant in clinical practice and post-graduate training, which will be described in detail in the next chapter. His contributions helped affirm and strengthen results.

Summary

The main research question for this study was: What are the lived experiences of emerging adults living with JOMD? A series of interviews generated rich descriptions of the phenomenon of living with this chronic, incurable, debilitating disease during the most unstable, unpredictable, stressful period of life. The researcher utilized a

phenomenological methodology for data analysis to describe the essence of the phenomenon. The results of the research study are described in the following chapter.

CHAPTER III

RESULTS

Chapter 3 presents the results of this phenomenological study examining the experiences of eight emerging adults living with a form of JOMD. First, demographic information of the participants is described, including ages, type of JOMD, age when diagnosed, employment and relationship status, highest level of education, current living situation, assistive devices and pets. Next, each participant is profiled in more detail. Finally, the themes that emerged from the data include are described and discussed.

Participants

The purpose of this study was to understand how eight emerging adults perceive and describe their experiences of living with juvenile onset MD. Participants' ages ranged from 23 to 29 years old. The sample included two males and six females; five identified as Caucasian, one Puerto Rican/Greek, one described herself as Muslim-American, and one German. Seven of the participants lived in the United States, and one lived in Germany. All participants agreed to participate in two 60–90 minute interviews describing their experiences of living with JOMD. It was ensured that the inclusion criteria (i.e., between the ages of 18 and 29; no known cognitive impairments or problems understanding English, had a form of JOMD; could recall memories of living both with and without the condition and; had been diagnosed with MD for at least one year prior to participation) were met. See Table 3 for demographic information.

Table 3

Demographic Information

Participant	Gender	Age	Nationality/race/ethnicity	Employment Status	Relationship Status	Highest Level of Education	Current Living Situation
Desmond	M	29	Caucasian	Disability	Single	Some college	Mom and 2 dogs
Lindsey	F	27	Caucasian	Full time	Married	Bachelor's Degree	Husband, infant son and dog
Sarah	F	28	Muslim American	Full time	Single	Bachelor's Degree	Parents, 3 sisters 1 brother
Lani	F	29	German	Disability	Engaged	Associates Degree	Fiancé
Mae	F	24	Caucasian	Disability	Separated	Some college	Mom and infant son & 4 dogs
Denise	F	23	Caucasian	Student	Single	Some college	Mom & sister
Jemma	F	25	Caucasian	Student; Part-time	Divorced	Some college	Parents and 1 dog
Sam	M	23	Puerto Rican/Greek	Student; Part-time	Single	Some college	Mom, maternal grandparents, 1 dog & 2 cats

The researcher gathered data about developmental milestones common among emerging adults, including employment status, highest level of education, relationship status, and current living situation, also listed in Table 3. Two participants were working full time, three were on disability pension, and three were students, two of which were also working part-time. All the participants had college experience; two with bachelor's degrees, one with an associate's degree, two currently enrolled in classes and three who have departed from their programs/colleges. Relationship statuses were mixed: single (four participants), engaged (one participant), married (one participant), married but separated (one participant) and divorced (one participant). Six of the eight participants were living with one or both parents, and two with their romantic partners.

Two forms of JOMD are represented in the sample: Seven of the participants had a form of LGMD and one had EDMD. Participants were diagnosed with JOMD between the ages of 5 and 21 years old. The years of experiences of those living with LGMD ranged from 4 to 20 years; the participant with EDMD had 9 years of experience. Assistive devices utilized because of effects of JOMD are listed. All participants began their experiences of living with JOMD with subtle or invisible physical differences. By emerging adulthood, seven of the eight participants in this sample required an assistive device of some kind, including a power wheelchair (three participants), a scooter (two participants), a cane (one participant) and a pacemaker (one participant). Two participants had utilized professional caregiving for assistance with ADLs. One has an oxygen machine he uses at night. See Table 4 for further information. Following the table, each participant is described in further detail.

Table 4

Experiences of Living With JOMD

Participant	Type of MD	Age diagnosed	Years living with	Assistive Devices
Desmond	LGMD2a	12	17	Power wheelchair & breathing machine
Lindsey	LGMD2d	13	14	None
Sarah	LGMD2c	8	20	Power wheelchair, leg braces; caregiver PRN
Lani	LGMD2i	5	24	Power wheelchair
Mae	LGMD2i	14	10	Scooter PRN
Denise	LGMD2a	12	11	Scooter PRN
Jemma	LGMD2b	21	4	Cane PRN
Sam	EDMD (X-linked)	14	9	Pacemaker

Desmond

Desmond is a 29-year-old Caucasian male living with his mom and two dogs, and was the first to participate in the study. Interview one was completed in the researcher's office, and the second in Desmond's home. He was an open and willing participant despite rarely talking in depth about his experiences of living with LGMD2a. He is in a power wheelchair most of the day, and uses a breathing machine at night. Social security is his means of income; he is unsure how he could ever work full-time. There is no one else in his known family history who has MD of any kind.

At first, his condition was hardly perceptible to him. He was diagnosed with LGMD2a at the age of 12. At the time, he did not see himself as different from his peers.

Desmond described the profound losses he encountered as LGMD progressed, and subsequent shame and guilt he felt about himself. He perceived his parents (especially his father) to respond with disappointment and/or anger for perceptible weakness, and he was bullied by peers in high school for falling easily and not being able to get up on his own. These reactions reinforced negative beliefs about himself and LGMD. Panic, anxiety, and depression set in when he was in high school and has lingered ever since. He is being treated pharmacologically for these conditions. Counseling services were utilized while he was in high school, but he did not find them helpful.

Desmond emphasized the mental fortitude it takes to live with LGMD. It has been difficult for him to persevere and feel hopeful when he has endured so many setbacks. He stated "after you just keep hitting brick wall after brick wall, it just starts beating you down and it's hard to keep getting back on your feet and keep trying to do it. I'm not getting anywhere . . . so why keep trying." The progressive and non-curable aspects of LGMD translate into constant hurdles that mentally and physically "beat" him down and make it difficult to have hope. He feels anxious about the future because he is unsure how severe LGMD will get. Currently, he states, "there aren't too many parts of life that (LGMD) doesn't weave itself into one way or another, so it is (my) life." He finds it difficult to enjoy his existence, and compares it to being stuck in purgatory.

Lindsey

The second person to be interviewed was Lindsey, a 27-year-old Caucasian woman residing with her husband, infant son, and pet dog. She works full time. She does not use any assistive devices. For convenience purposes, Lindsey preferred to

complete the interviews via telephone. She is the only known person with MD in her family tree.

Lindsey began her narrative by sharing a family history of athletic achievement. When she was a young child, she participated in and excelled at sports, though she recalled being inordinately sore afterwards. At age 13 she inexplicably declined in her cross-country and track times/rankings. When she pushed herself to run faster she lost control and fell. Her parents noticed these differences before she did, but when they pointed it out to her it resonated with how she had been feeling. During the diagnostic process, she had to cease all sports (to reduce the level of the enzyme creatine kinase found in her blood that rose from skeletal muscle injury from MD) and was being bullied by her classmates for her hypertrophic calf muscles. It was a difficult time in her life.

Lindsey highlighted the challenges she faces living with LGMD, notably a growing awareness of her limitations brought about by travelling for business and motherhood. She is at a place in her progression where she appears largely unaffected, but has significant trouble walking up stairs and rising from a seated position. Inclines are difficult. These tasks are problematic and at times impossible, but not severe enough to necessitate an assistive device. She worries about climbing stairs in new social situations. She feels more pressure to explain her physical disability to strangers or new acquaintances to avoid negative judgment and possible embarrassment, since it catches people off guard that she has such severe physical struggles being only 27 years old. She reported feeling uncomfortable talking about LGMD, and rarely does, to anyone except her husband and immediate family, seeing it as a personal vulnerability.

She feels inferior because of her limitations, stating: To live with LGMD is to be humbled time and time again. She phrases it as follows:

> Sometimes, I feel a little bit less of a mom or a wife, because I want to do things and I wonder if I am holding (my husband) back from doing stuff. That's tough. So, I think the hardest part is being held back in those different roles you play in your life.

She finds it difficult to not compare herself to others and feel self-conscious and frustrated that little things, like getting up from the floor and picking up a child, are difficult or impossible for her. No matter how much personal or career success she has, she still feels substandard because she cannot perform these everyday tasks.

Sarah

Sarah is a 28-year-old Muslim-American female. She has a bachelor's degree and is working full time as a graphic designer. She lives in Northeast Ohio with her parents, three sisters, and one brother. She has a cousin with an unknown form of MD. Sarah requested a phone interview. The second interview was completed but, unfortunately, not recorded, so only the first interview transcript was utilized for data analysis. Sarah utilizes a power wheelchair, drives an adapted vehicle, and recently hired and fired her first caregiver when she discovered she was stealing her jewelry.

She describes living with LGMD2c to be filled with frequent ups and downs, good days and bad days. When she has a bad day, it is usually because she had to wait a long time for someone to help her with a personal task, such as use the restroom. Sarah articulates, "I'd like to be able to not have to wait anymore. Just keep going. To be able

to get up when I want instead of saying, okay now I have to wait." Equipment failures throw off her routine, and she is forced into a stressful adjustment process. She stated, "Then, eventually, I'll probably get used to it being broken. Then, when it does come back, I'll . . . have to learn again." She reflected on the difficulty of decreasing independence over time.

Sarah reported being very self-conscious during her pre-teen and teenage years. Every fall lowered her self-esteem. Eventually she wrote letters to her peers explaining her disability, and she noticed an increase in kindness and acceptance. While she is doubtful she will ever experience marriage, motherhood, or independent living, she is more confident and self-aware because of living with LGMD. She reported, "I learned how to adapt to the world around me, how to make it my world versus other people's world." Transitioning into a wheelchair increased her feelings of power, a surprise side effect of a negatively anticipated event in her life, occurring during her college career. Sarah reported no history of mental health treatment.

Lani

Lani is a 27-year-old female residing with her fiancé in Germany. The researcher completed both interviews via Skype. She is currently on disability, but previously earned an associate's degree and worked as a secretary in a public school. She utilizes a power wheelchair and a professional caregiver, especially when her fiancé is out of town. She was an enthusiastic participant, as evidenced by disclosure of personal struggles in mental health and counseling treatment. Where she resides, in Germany, she has a network of others with neuromuscular disease with whom she exchanges, giving and

receiving, support. English is her third language, German and French being her native tongues. Nonetheless, she provided rich descriptions of her experiences.

For Lani, living with JOMD began at the age of five when she was diagnosed with "In-specific MD." However, it wasn't until elementary school gym class and inability to keep up with her peers running (18 months later) that she became conscious of her condition. She felt like the "black swan," aware that she was different, and wanted to understand more. She vividly remembered learning of the progressive, degenerative, non-curable nature of her disease, because it is when she lost her faith in God:

> When I got to know that there is nothing that you can do . . . I decided for myself that there can't be a God, because if there was God and Jesus then I wouldn't have this disease . . . Why would He do this? It was kind of terrifying for the little me. And it (hasn't) changed so far—I still don't believe.

She acted out emotionally by externalizing anger and sadness; she bullied other children, stole small items, and frequently threw tantrums. Over time her symptoms improved, accelerated by an accepting friend group.

Lani emphasized the emotional aspects of living with a physical condition and her personal experiences of loss.

> The most difficult aspect is to be fully and constantly aware of losing all your body functions. Of being aware of the progressing and to really see how my body and me turn into a puppet . . . a marionette. It's my feeling . . . I am the observer of how my body is always getting weaker and weaker and I can't do a thing about

> it . . . The awareness and that I can't run away . . . I stay conscious, my whole life I stay conscious about the wasting away.

She described debilitating panic attacks that began in her early 20s. At the onset, she thought she was having MD related heart failure. Through counseling, rest and self-care, she recognized how fearful she was of integrating MD into her life and utilizing a power wheelchair. Her overall emotional state has significantly improved since her early 20s, but she described an underlying depression and sense of hopelessness that lingered. She stated,

> There are days where I really tell myself, hey, you're not worth a thing. You can go and throw yourself somewhere and then it would be over. You wouldn't have to struggle with life because sometimes it can be really tough.

Over time, she has learned to accept uncertainties and feelings of discomfort that come with LGMD, to the point that she is able to travel and socialize, activities that initial cause anxiety but, upon successful completion, significantly improve her sense of wellbeing. She travelled to Cape Towne, South Africa, between interviews.

Mae

Mae is a 24-year-old Caucasian female, diagnosed with LGMD2i and residing, at the time the phone interviews were completed, with her 57-year-old mother, infant son, and four dogs. She is divorced from her husband. She has been receiving social security since high school. Mae hoped to, someday, work in the mental health field, but departed from higher education due to mobility restrictions (e.g., difficulty getting out of desks by herself). She stated,

> I was motivated after (high) school. I felt like I was going to go to college and do a good job, but after things started getting harder and harder I just kinda gave up on myself . . . I lost a lot of motivation and hope.

Her mom aids with ADLs and childcare.

Mae describes living with LGMD as being caught between two worlds—the able and the disabled—not perceiving herself to fit in either one. She can walk, but it is slow, unsteady and exhausting. She has a lot of trouble getting up from a seated position. Her disease progression became most apparent to her when she fell – there are times she cannot get up and is stuck on the floor until someone can help her. She reflects:

> I get really frustrated at times because I used to be able to help myself up or push off things, but when I couldn't, I could still roll over and get on my knees and help people a little bit more and then recently like the past year or two, if I fall and I'm in position I can't move anymore. I can't turn over or get on my knees. I can't push myself up on my arms or anything. I just kinda lay there and that affects me a lot. I get really frustrated with that.

She feels anxious at the thought of getting a power wheelchair as she does not want to depend on equipment, and does not want to give up driving. While she yearns to go places by herself, she doesn't, out of fear that she will fall and need help getting up. Her self-esteem has plummeted over the past two years. She feels inferior when she compares herself to others, and has grown to avoid social activities. Her physical, mental, and emotional declines impacted her marriage, which recently ended when she learned of her husband's infidelity.

Every day Mae feels frustrated and depressed; about every two weeks she emotionally breaks down, feeling frustrated and hopeless her life will improve. Sometimes she feels depressed for several days after a fall, grieving what she cannot do and questioning the point of living when she will eventually have to rely on others. Her PCP prescribed an anti-depressant medication a few weeks before the first interview, and she would like therapy to "learn how to like (herself) again" but has yet to initiate services. She feels lost and unhappy, unsure how to get out of this mental place. It gives her comfort to believe that, when she dies, she will go to heaven and be free from LGMD. She reflected on the mental strength it takes to live with this condition. She tells herself to focus on the positive and accept her limitations, but the inability to do so reinforces negative feelings about herself.

Denise

Denise is one of the youngest participants, interviewed at age 23. She lives with her mother, sister, and pet dog. She described her ethnicity as Caucasian, and was, at the time of the interviews, nearing graduation with her Bachelor's in Communication. She occasionally utilizes a scooter for long distances, especially outdoors, but otherwise her disability is hard to detect. Denise completed both interviews over the phone.

Denise spoke at length about the years of denial and emotional repression she experienced attempting to hide her symptoms of LGMD, both from herself and others. She was diagnosed at age 12—after noticing muscle weakness and toe walking since 1st grade—but was told by her pediatrician that she would eventually outgrow the problems. Within 6 months, she learned more about LGMD, and felt ashamed and embarrassed of

it. For the next six years, or from 12 to 18 years old, she had a very hard time talking about the condition. She felt anxious about her symptoms being obvious to others, did not know how to talk about it, and was embarrassed to ask for help. Her parents' reactions were memorable in her experience; her dad would virtually ignore her struggles and tell her to "keep exercising" and her mom would get outwardly distressed and loud if she fell. In hindsight, she realized these responses exacerbated anxiety about living with LGMD. She saw a therapist in 11th grade, but stopped because she did not like it. By her first year of college, she could no longer deny that she had gotten progressively weaker and would need some type of assistance—a recognition that brought great sadness, anxiety, and avoidance behavior. She reported:

> It definitely felt overwhelming, and I feel like I did not deal with it for most of that time from 12 to 18 so I think it catches up to you and you realize like oh, I have to deal with it and not just ignore it.

Over the course of the last few years, Denise has gotten more comfortable telling others she has LGMD, but feels vulnerable "giving away a big part of (her) private life right away." Establishing a supportive social group has improved her confidence. She expressed gratitude for one friend who advocated for Denise's needs, when she was not yet ready to do so, and role-modeled how to speak assertively about LGMD and the need for accommodations. While her emotional distress has improved, she continues to feel upset and left out at times. She is nervous that people will reject her; at night, her mind races, making it difficult to fall asleep. She is worried about graduating college and finding a job. She is pessimistic about someone being willing to marry her given her

condition. It is difficult to feel hopeful about the future. She has wanted counseling services over the last 18 months, but has yet to go. She reported that it would be helpful to talk about her thoughts and feelings without overwhelming her family members. Participating in the interviews helped her become more aware of where her feelings were coming from, which allowed her to dispute automatic thoughts, thus taking away their power.

Jemma

Jemma is a 25-year-old divorced Caucasian female, living in rural Pennsylvania with her parents and pet dog. She works part time in retail, and is also a student. For convenience purposes, Jemma completed both interviews over the phone.

Jemma was diagnosed with LGMD2b at the age of 21, nearly a decade after she first began noticing symptoms (e.g., being a slow runner) in the 7th grade. Her primary care physician assured her that the muscle pain and weakness was a result of "growing pains," and with supportive shoes and diligent exercise, she would physically catch up to her peers. As her physicality inexplicably declined, she grew frustrated and wondered if she was lazy, mentally crazy, or weak. In high school, she avoided stairs and physically challenging tasks. Gym class and climbing the stairs in her school were tasks she came to loath and dread, both because of the effort and the possibility of others detecting her disability, which she did not know how to explain.

After high school her symptoms got worse, and her parents encouraged her to pursue another medical opinion. Finally, at the age of 21, she was diagnosed. She was

initially relieved to know she was not to blame for the weakness and pain; it gave her peace of mind to know it was not "in her head" and there was a medical explanation.

She is now at a point in her progression where she can get around without any assistance, but struggles to have the energy to do everything she wants to do. Working more than four of five days in a row will exhaust her. If there are activities she wants to do in two consecutive days, she picks one, because her muscles will not have the ability to do both. Along with fatigue, she experiences intense pain in her legs and back that makes it hard to relax and sleep sometimes. She occasionally takes a prescription pain killer to dull the ache, so she can rest. Nonetheless, she does not like help of any kind. Doing things herself eases anxiety about her limitations and fear of depending on others in the future. She is determined to keep her mobility as much as she can and not rely on anyone or anything. She worries how much worse she will get, and fears the losses will devastate her.

Sam

The last participant to be interviewed for the study was Sam, a 23-year-old single male living with his mother and maternal grandparents. He described his ethnicity as Puerto Rican and Greek. He worked part time and had some college experience, but dropped out of several career programs (police, fire, automotive) because of unexpected EDMD related issues and is currently unsure what occupation to pursue. He was initially reluctant to participate in the study, but with strong encouragement from his mother (who saw the study advertisement on Facebook), he completed both interviews over the phone.

Upon conclusion, he expressed benefit and cathartic relief from sharing his story. He has a cousin and grandfather with EDMD.

Sam was shocked by his diagnosis of EDMD at age 14. Unlike with the other participants, there were no detectable signs of muscle weakness that led him to seek medical consultation. It was his grandfather being diagnosed with EDMD (after decades of misdiagnosis) and questioning the hereditary nature that got everyone in the family to undergo genetic testing. He was shocked and in disbelief upon learning he had EDMD. He was told to stop intense sports, like BMX bike riding and skateboarding, because it could accelerate EDMD related problems. A year later he had to have surgery to implant a cardiac pacemaker, and he remembers waking up from surgery furious and fearful he would die unexpectedly. He commented on how difficult this was for his family:

> I think everyone in the house got hit. Everyone in my family got hit along the way. There was a lot of anger, I know that. Everyone was always yelling at each other, screaming at each other, and there was a lot of hostility in the house at the time. I don't think anyone knew how to deal with it.

The next year of his life he describes as a "hole in time;" he has virtually no memory of follow up doctor visits; his school performance and attendance declined; it is as if he detached from reality to block out trauma. He was forced to go to therapy, but was not engaged or interested at the time. When he talked to his family about his feelings, he felt invalidated and would be lectured on ways to improve his attitude.

Sam emphasized the mental and emotional aspects of EDMD that have left him "mentally shattered" over the last two years. He had never disclosed how much he has

struggled with repressed feelings of inferiority, grief, anger, and anxiety about when and how EDMD will progress in the future. He has tried to cope on his own, but realized he needs mental support. He reported:

> This condition isn't about the physical problems, it's the mental problems that come with it. When you get diagnosed with something and people look at you, they say, oh you can stand up straight, you can walk on your own, but not all conditions affect your body, they can affect your state of mind and that's a lot harder to recover from than a broken leg.

While his physical limitations include restrictions on airplane travel and metal detectors (due to the pacemaker), he is reminded of EDMD "over and over again" when he feels muscle pain or needs accommodations; then it's like hitting a "brick wall" that angers and upsets him, reminding him of the condition. Sam described feeling taunted by symptoms of EDMD that depress him when he was otherwise feeling happy and/or successful.

> It's something that pops up whenever you have a good moment and then it pulls me back down . . . I have my goal but this is its own entity that has the ability to kick me . . . and then I have to try to get back up again.

It is difficult to enjoy what he has now, because he fears it will get difficult in years to come.

Summary of Participants

This sample represented two forms of JOMD – LGMD (n = 7) and EDMD (*n* = 1). Of the eight individuals interviewed, six were Caucasian, one Puerto Rican/Greek,

and one Muslim-American. Six females and two males comprised the sample. The average age of diagnosis was 12.4. The mean years of living with JOMD was 13.6. None of the participants were living alone at the time of the interviews. All but one participant utilized some form of assistive device because of living with JOMD.

All participants thoughtfully and elaborately responded to the research question. The researcher, also having a form of LGMD, likely aided in disclosure on behalf of the participants. For about half of them it was the first time they had talked to another person with JOMD. Nonetheless, each wanted to contribute as a way of helping others who also live with JOMD. All participants maintained contact throughout the research process, and were interested in learning the results.

Experiences of Emerging Adults With JOMD

This study utilized a phenomenological framework articulated upon by Giorgi (1997, 2009) to reveal the psychological experience of living with JOMD, specifically in this case LGMD and EDMD. The phenomenon of emerging adults living with JOMD was investigated to reveal the essential components of the experience. The goal was to highlight the mental and emotional process of living with JOMD.

The research question guiding this study was, "How would you describe your experience of living with your condition, beginning with your first memories up until now?" Sub-questions were utilized to gather more information, and were as follows: (a) What aspects (being diagnosed, loss of ability, etc.), occurrences, or people connected to the experience stand out to you? (b) How does having MD affect you and your life? How has it changed you? (c) How do you believe having your condition has affected

significant others in your life? (d) What is the most difficult aspect of having this diagnosis as an emerging adult? (e) What, if anything, do you believe could be helpful to you to better cope with your condition? (f) How do you expect your condition will impact your future? (g) How did you get more information about your diagnosis? (h) Have you shared all that is significant about your experiences of living with this diagnosis?

The five-step approach to uncovering the essential elements of living with JOMD was carried out systematically. First, the transcripts were read holistically. Second, the transcript was broken down into meaning units, or smaller units of text that describe an aspect of living with JOMD. Next, the meaning units were transformed in a three-step process from verbatim statements into scientific expressions via "transformation chart." Fourth, the data from each participant's 3rd transformation was organized and expressed in an "Integrated Narrative" (see Appendix N for an example). Finally, constructs shared by participants were cross-referenced and elaborated upon to form themes.

While the story of each participant was unique, data analysis revealed similarities across participants. Five themes emerged from this study after cross-referencing each Integrated Narrative to describe the experience of these eight emerging adults living with JOMD. Ultimately, the research question that led to the most substantial information and theme development was the first: "How would you describe your experience of living with your condition, beginning with the first memories up until now," which essentially examines first noticeable symptoms, diagnosis, and progression. Participants had the most to say about this topic. When asked, "how do you expect your condition will impact your future?" the response was consistent. Subtleties of data gathered from

sub-questions are incorporated into five main themes that represent the "essential elements" of this specific, yet broad, experience. Giorgi (1997, 2009) suggested each theme be a pillar of the experience—that without it the structure would collapse. On the other hand, too many themes could dilute what makes living with JOMD different from other juvenile onset chronic diseases.

The first theme emerging from this study is that JOMD expands into consciousness with continued loss, or becomes more intrusive in life over time. Second, the stress of experiencing increased visibility; participants described difficulties as progression made symptoms of JOMD more noticeable to themselves and others. The third theme, self-other justification of JOMD, depicts how individuals make meaning of JOMD symptoms and how they navigate telling others about the illness. In the fourth theme, evolution of disease, evolution of negative emotion, adverse psychological aspects because of JOMD are coming to the fore. This is the only theme broken down into sub-themes, or four mental states that accompany JOMD, mainly suppression, anxiety, depression and fatigue. Fifth, fear of the future captured the angst of living with a progressive, degenerative, incurable illness. The remainder of this chapter is dedicated to explaining the research findings in, detail.

Theme 1: JOMD Expands Into Consciousness With Continued Loss

The first theme was that JOMD is a condition that grows into consciousness over time as affected individuals experience new and evolving limitations. It begins as relatively non-intrusive phenomenon and is thus given little thought or attention; by emerging adulthood, symptoms impaired everyday functioning for participants. Simply

put, the amount of mental space occupied by JOMD expands over time through repeated experience of loss and limitations.

In the early years of living with JOMD, participants did not often consider the diagnosis (if they had one). Awareness of its existence came into being through losses—either pre-mature muscle weakness, physical pain, or limitations in functionality. Otherwise, it is forgotten. Sam describes his early experience of feeling unaffected, then remembering he lived with EDMD:

> The first thought that came to my mind back then, like 10 years ago or whatever it was . . . surreal. It didn't really make any sense. It still doesn't. And all throughout the time it's just been not really making sense. I go and try and do something and then I realize can't really do that, or I have to get (doctor's) approval for that. So, it's been an up and down thing where you forget about it and then you do something and it's like—ah, it's there.

The initial nonintrusive nature is echoed by a participant with LGMD (Mae): "When I was younger, it was not very hard. I just noticed I was a little different, mainly in gym class, and . . . I was just very slow. I was always the slowest person." Living with JOMD was not considered a big part of life in the beginning. The condition is present but not given much thought about in the early years.

Several participants describe the experience of learning that they could not stand up independently as another instance of realization and growth of JOMD into consciousness. Standing up from a seated position evolves to become unpredictably impossible and may require help from an able-bodied person. This loss experience

significantly increased the number of times, the regularity with which one thought about living with JOMD. Not being able to consistently stand up on her own caused such stress for Mae that she decided to stop going to college, to avoid having to ask for help.

> I couldn't get out of the seats. And that made me feel really bad because I enjoyed going to school and getting out, but I was just so embarrassed if people stared at me . . . And if I got stuck and I would have to ask for help, so I stopped going for a couple years.

Desmond remembers his first experience of unexpectedly needing help as a turning point in his mind that awakened him to a new and troubling aspect of LGMD.

> I remember the movie, it was the scene at the end, because I was sitting in a movie theatre seat and I knew I couldn't get back up myself. I knew I needed someone to help me out of it and that was, I guess, a little bit of claustrophobia; not that I have claustrophobia, but a kind of it.

Growth of JOMD into consciousness occurred during a change in residency when Mae learned she could no longer get up off the toilet without assistance. Having to consider when to use the restroom, so she would be able to get up, became a new part of her everyday routine.

> As long as I had something in front of me, I could push off my knees and then grab onto something in front of me and get off the toilet that way. I didn't need help at home but then we moved into a new house here and I still had the toilet seat but there is nothing in front of me, so I couldn't grab anything, so I had to stop doing that and I just needed to be picked up from then on.

Unexpectedly needing help, especially on the toilet, is perceived as a loss and causes patients to become more aware of avoiding, if possible, a repeat of the experience. Lani thought much more about locking a bathroom stall door after it took a long time for a friend to get to her when, unexpectedly, she found herself too weak to get up on her own.

> I crashed on the wall, I fell down on the floor, and I couldn't get up. Then I also had to cry out for my friend . . . and she had to open the lock with a coin because I locked it. And, from this day on, I don't lock when I use the toilet . . . because I always think this situation could reoccur . . . This was a changing point also because from then on . . . when I went to the bathroom I thought hey maybe now I won't get up by myself. I always have to figure out a way to alert someone else to help me if I really should need help.

As JOMD progresses and symptoms worsen, adaptations grow and there are more circumstances to consider, such as how to complete activities of daily living when the equipment one relies on changes or stops working. Small changes can throw off one's ability to manage tasks of daily independent living, and energy is expended to solve the problem. For example, Sarah describes how a seemingly simple deflation and replacement of her wheelchair cushion, threw off her routine and caused her to reflect on living with JOMD.

> Last week my wheelchair cushion stopped working. I guess it deflated and it's making me uncomfortable because I have to adapt until I get a new one in. It just annoys me that I have to adapt to certain things. I rely on that chair and I have to wait a certain amount of time in order for it to get fixed . . . Then eventually I'll

> probably get used to it being broken. Then when it does come back, I'll be like great, I have to learn again.

A wheelchair cushion that is a few centimeters different from the one she is used to is an obstacle that requires attention. Living with JOMD is a never-ending cycle of loss and adaptation. What begins as a condition that hardly comes to mind in day-to-day life evolves to occupy more and more of an affected individual's consciousness by emerging adulthood. For those who are more severely physically disabled, the awareness of living with JOMD is nearly constant, as described by Lani:

> I notice it (LGMD) when I open my eyes and when I want to get out of bed. Then I am already confronted with what is LGMD. But obviously, I don't talk about it every day. But in my daily life it's present from the morning until when I got back to bed in the evening. Because I'm so severely affected that I'm not able to forget it anymore.

Desmond is at a similar point in his progression of LGMD (full-time power wheelchair user, capable of some ambulation) and shares similar thoughts about how his condition has expanded into his consciousness over time. It seemed inevitable that each participant would come to realize more and more exactly what it means to live with JOMD. Desmond states it succinctly:

> It is on my mind a lot . . . always . . . It is everything. Because it is progressive, you don't think about it at first. But then, ya know, down the line things just keep getting worse and worse and worse so you think about it more and more and more.

As one is more severely limited by the effects of JOMD and confronted with limitations, participants increased the frequency of cognition about the condition. Mae shared the continual self-talk she has about JOMD as she lives her everyday life.

> I would say that it is always on my mind because every day I'm slow I'll be like dang, I wish I could walk faster—this is annoying . . . I wish I could get this, I wish I could get there faster, I wish I could bend down and pick up that thing I just dropped. Stuff like that so it's always on my mind. Like I never have a chance to feel normal.

Denise described her growing awareness of JOMD in day-to-day life. Each loss she encountered added to a list of considerations for leaving her home.

> If I go somewhere, I need to think through everything, like where is the bathroom? Am I going to be able to access it? Is the toilet really short? And is that going to be hard for me? Are there steps? Do the steps have a railing? Are the chairs . . . I don't know . . . a certain way that makes it difficult (low to the ground)?

JOMD is a condition that grows into consciousness over time as an affected individual encounters various and continual losses.

Theme 2: The Stress of Experiencing Increased Visibility

JOMD is a condition that grows more visible to the affected individual and others over time. The virtually undetectable nature of JOMD was reported as both a positive and negative aspect. On the one hand, the imperceptibility protects individuals from immediate identification as a person with a disability, but on the other hand it masks how

mentally burdensome symptoms can be and makes inevitable deviations from the norm embarrassing and upsetting. The visibility barrier was an important part in understanding the experiences of living with JOMD as an emerging adult, because self- or third party detection of symptoms are both described as stressful. The barrier is ultimately broken by events that showcase any abnormality in functioning, or occurrence requiring assistance from others. The visibility of JOMD increases substantially, to oneself and others, by the addition of an assistive device or technical aid. Up until this point, no obvious signs of disability are presenting themselves to the public.

Life with JOMD begins mostly or totally "normal," or without significant limitation. During elementary and middle school, symptoms spring up, which are described as embarrassing and frustrating. Tasks requiring intense physical movement, such as running or squatting, drew attention to the presence of JOMD. Hypertrophic calf muscles are another unwanted visible symptom described for those with LGMD.

Reactions from parents and primary caregivers played a role in how JOMD is interpreted and later expressed or repressed for this sample. Those who had a helpful, knowledgeable, support system had an easier time communicating and adapting to physical changes. By contrast, parents who lacked knowledge about JOMD or overreacted to symptoms, increased fear in the participant and repression of negative feelings. Desmond described how his father's response intensified the loss of learning he could no longer run:

> I remember it was winter, and you are inside for the winter, and then when it started becoming spring I started going outside and I realized I couldn't run. And

> then my dad, well this was traumatic, but my dad made me try to run up and down the hall and he's like, "come on, you gotta try and run" and I'm like, "I can't. I forgot. It's not working." I did not feel good. It was, it was a very traumatic experience . . . I was in the mentality, and still in the mentality of kinda being normal, of fitting in with everyone, and ya know, this made me an outsider.

As Denise got physically weaker, she did not talk to anyone about changes she was making to carry on as normal, but felt shame when she was "caught" doing something differently. She described feeling negatively judged by her mother when she sat down to get dressed:

> I remember I sat down to put on pants and I think my mom was like, "I saw you do that." I was . . . trying to hide that I was sitting down, and I don't think she realized that I felt like I was doing a bad thing. I don't know, I guess maybe she was like, I wonder why she is trying to hide these things from me but I felt like I had to. It felt like I was doing something bad, I guess, like it was my fault.

Third party attention, paid to symptoms of JOMD, intensifies the patient's internal realization that JOMD is indeed progressing and becoming more noticeable to others, which in itself adds stress and anxiety. It increased patients' cognitive dissonance between seeing oneself as normal and healthy versus abnormal and impaired. Adolescents and emerging adults with JOMD discover symptoms alongside others who might be there to notice or point them out. As long as the affected individual can navigate most tasks independently, no help is needed or wanted. Attention on them about symptoms or limitations of JOMD is often uncomfortable and sets them apart from

feeling normal−or accentuating a difference that one wishes did not exist but can do nothing about−and tended to elicit feelings of shame and doubt.

Participants attempted to minimize the visibility of JOMD, or disguise initial symptoms from others. Great efforts were made to appear unaffected, as Denise illustrated in the following quote:

> I would always be really nervous about standing up from a chair, and so I would always just think okay, how can I get up? How can I get up? What should I do? Should I wait until everyone else leaves so no one has to see me?

Appearing unaffected in the early years of living with JOMD is not usually difficult, if one avoids strenuous physical activity. Fitting in was important, and participants rarely talked about their condition to anyone. Participants described going to considerable length to avoid others detecting JOMD symptoms out of fear of rejection. For example, Desmond made up excuses to avoid a gravel path that connected the school buildings because the unsteady ground could easily throw him off balance and lead to an embarrassing fall. He described making sacrifices for a better social life, to protect his secret of living with LGMD:

> In high school, I would have a girl that would . . . be like, "Hey, you want to come to my locker with me?" And at my high school there was this short cut you could take to go outside . . . like this stone path that gets you from building to building, or you take the long path (that was paved) so everyone would travel along this little (gravel) path . . . I would have to sit there and say no, because at that time I am not giving an explanation for it . . . and now we are not talking.

One source of anxiety included fears of rejection based on having JOMD. Worry about being judged by others is common. Mae reported:

> Growing up I was a very anxious person. I didn't like attention and I don't like people staring at me or asking for help, but as I got older, I have gotten better at not caring what people think so much.

Denise describes: "It's a deeper issue of what is going to come of it and are people going to reject me or am I going to feel badly about myself as a result?"

The visibility of JOMD to peers increases fear and inferiority in participants as it accentuates a difference, or limitation, that is perceived as weak. Falls, especially at school in front of peers, were particularly distressing and embarrassing due to the visibility of physical feebleness, which sometimes brought negative peer attention. Desmond expressed emotional pain from recalling the reaction of his male counterparts when he fell in the high school hallway.

> I would fall in high school. I remember, I fell against the lockers and I just waited until everybody left and I went and there was like a chair all the way across the room and I had to crawl towards the chair to help myself up and then these two jocks came by and they were like, "God gave you legs for a reason," so that doesn't help things. Or, when, I also would trip in the hall and everybody is walking by and they would squirt water bottles in my face and laugh at me.

Emerging adults with JOMD struggle socially and emotionally as JOMD becomes more difficult to camouflage while completing everyday tasks. At the same time, the invisibility of JOMD to the public was also a challenge reported. Applications for

accommodations such as handicap parking and receiving social security benefits were met with disbelief and scrutiny. Participants reported stigma and negative judgments from the public for not being more obviously disabled. Jemma described perceived stigma when she tells someone she is on disability: "It's irritating because people look at me and they think at my age, why aren't you working, why don't you have a full-time job? I don't look like there is anything different about me."

At other times, the public is caught off guard by the muscle weakness and an intrusive or hurtful comment is made. Lindsey reported feeling nervous when her condition becomes visible to strangers and she gets stared at or questioned.

> The most challenging for me and my young adult life is when I can't do something, people like "what the hell are you doing?" They . . . judge you. I'm self-conscious about it because I know what I can and can't do but how do you break that conversation threshold with somebody to tell them what your limitations are when you just meet or when it's in a business sense.

The visibility barrier both helps and hurts those with JOMD. It allows those affected to live part of life feeling "normal" and it can, in the beginning, hide symptoms from others, allowing the patient not to disclose the chronic illness. Over time, hiding one's limitation is stressful and impossible. Reactions from parents and peers play an important role in how one responds to the growing visibility and progression.

Theme 3: Self-Other Justification of JOMD

The theme of "self-other justification" describes the private psychological process of rationalizing limitations, or how progression was interpreted by participants at

different points in time and physical ability. Though participants knew of their diagnosis, many struggled to understand or explain symptoms to themselves and others. This then features, alongside increased visibility of JOMD, attention from others who notice a limp or question the legitimacy of accommodations. At this point participants felt obligated to provide some rationale to onlookers, which was challenging and stressful. The experiences of explaining JOMD and justifying its effects to oneself and others are described next.

Participants show a high degree of consensus on initial justifications to themselves. It comes with the realization of the catastrophic nature of their condition, paired with a sense of inferiority because of the limitations it imposes on them. Affected individuals personalized what they could not do, perceiving themselves as lazy and/or inadequate for experiencing muscle weakness or needing help. It can be difficult to justify to oneself why simple tasks are difficult without feeling bad about themselves or projecting worries about future problems. Lindsey describes her automatic response to a limitation as follows:

> The default is that I do have that negative thought and it is kind of doomsday . . . it's so hard to change my default setting, (which) is "things are getting worse, everybody is judging me, things are getting worse, I used to be able to do this." My thoughts go "bing bing bing!" and then I have to coach myself through . . . it is an emotional thing at first and then it becomes logically thinking through it in a productive way.

Over time, accepting help became easier for participants. For example, Lindsey shared her automatic thoughts about having to ask a stranger to hold her infant son so she could use both arms to get out of a chair: "I'm so worried . . . I'm a bad mom because I can't stand up while holding my kid." Feelings of inadequacy were paired with learning of a limitation.

Asking for help or using a technical aid was especially difficult to justify to oneself, and approached with ambivalence. Participants describe taking pride in unassisted living for as much and as long as possible and perceived adaptions/accommodations as "giving up." Jemma reported intense muscle pain from difficult physical activities like carrying her laundry upstairs independently, but felt determined to avoid relying on needing help from anyone. Others were reluctant to make changes because, to them, holding out was a way to confront the fact that JOMD was progressing and would not be getting better. The costs of avoiding help or assistive devices were consistently paired with reduced social and occupational involvement.

Justifying symptoms to others (e.g., explaining the reason for taking an elevator, having trouble standing up, or falling) was consistently an issue for emerging adults with JOMD. It was difficult to tell others about the condition, not just because the public has little awareness of MD, let alone the Limb-Girdle or Emery-Dreifuss types of the disease, but also because participants felt embarrassed by their limitations and for needing help for a problem that is not visually obvious. Lindsey described the tension she experienced when others assume she is not disabled and expect her to climb stairs, a task she can perform but not easily or quickly:

> I'm standing at the bottom of a flight of stairs in front of people that I just met in a business sense and go, "Hey, is there an elevator around?'' . . . I don't approach (the stairs) and they go yeah . . . But they're like, "Well you don't want the exercise?" Then it traces to that now I need an explanation.

She then must decide how to explain her search for an elevator. Participants described feeling negatively judged by others causing them to want to justify why a task is difficult for them, but also not wanting to be a spokesperson for JOMD. When Sam told others that being diagnosed with EDMD ended his dreams of becoming a pilot, firefighter, and mechanic, he perceived others as skeptical and quick to offer unwanted advice.

> Then you have people coming up to you and telling you to go to school and do this and do that and you look like this so you should be able to do this and then you have to explain to them all over again the condition and they just stand there and look at you and are like but you look physically fine. It's like, you didn't listen to anything I just said.

Several participants reported envy of those with more commonly known conditions, such as cancer or multiple sclerosis, where no justification was needed to receive sympathy and understanding from others.

Aside from the public at large, justification to parents was another obstacle. When participants did express feelings of anger or sadness about falling or needing surgery, parents would sometimes invalidate such distress by telling their affected child to "not think that way" or "not to worry" about such things. It was frustrating to have to justify to one's parents why seemingly small things, like going to the neurologist, or a

negative comment from a peer about physical weakness, could be a devastating experience.

Dating posed another difficult conflict, as to how and when to disclose to a possible romantic partner that you are inflicted with JOMD and what that means. For example, Mae was using an online dating platform and in a sense felt like she was being deceptive because her disability is not visible in the photographs of her. She felt compelled to disclose her condition early on for the romance to evolve.

> I look good in my pictures and stuff and I try to talk to them normally and then I get scared—like when am I going to tell them this and are they going to accept it and keep talking to me? And that's a really scary feeling.

Sometimes participants lied to avoid a long explanation or rejection from someone they just met. The "tell or not tell" conflict created extra stress in social situations. Living with JOMD is a private event that is difficult to disclose, especially to romantic partners interested in a long-term commitment. Justification of JOMD to self and others was a repeating theme across participants in this study of emerging adults.

Theme 4: Evolution of Disease, Evolution of Negative Emotion

When asked, "How does having MD affect you and your life? How has it changed you?" participants shared stories of their condition impacting emotional as well as physical health. Emerging adults with JOMD reported different mindsets over time as they acquired more experience of living with the illness.

The theme "evolution of disease, evolution of negative" emotion was reflected in each narrative in varying and complex ways. As the researcher analyzed the data, four

sub-themes emerged specifying different mindsets central to the progression of the disease: (a) JOMD related suppression, (b) JOMD related anxiety, (c) JOMD related depression, and (d) JOMD related fatigue. Each is described further in sections to follow. Not every participant experienced each of the affective states portrayed in this section, thus sub-themes were created that comprise the descriptions of how living with JOMD negatively impacted emotional health.

JOMD related suppression. JOMD was described as a difficult condition to accept, especially during adolescence and early emerging adulthood, and participants described suppressing thoughts of their illness initially. While there was an awareness of the diagnosis, participants did not take help or acknowledge their true feelings about how JOMD symptoms impacted them emotionally. Ignoring feelings and thoughts related to JOMD served as defense from confronting the muscle or cardiac deterioration, as Lani described:

> Ignoring is a protection for us, I think. It's like I protected myself from the confrontation with the progression . . . because progression is never nice. Never. Because you always have to say goodbye to some functions. It's hard sometimes.

There was a tendency to shield oneself from the reality that the symptoms had progressed or that they would reach a point where the patient would need help. Jemma describes:

> I feel like I'll cross that bridge when I get there, but I don't want it to discourage me now. I don't want to think about any problems yet. I'll take it as it comes . . . I don't want it to define how I live my life.

Ignoring or suppressing works until the disability gets to a point where it can no longer be overlooked, a tipping point Denise described as distressing.

> It definitely felt overwhelming, and I feel like I . . . didn't really deal with it for most of that time from 12 to 18, so like I think like it kinda catches up to you and you realize like oh, I have to deal with it and not just ignore it.

The first few years after being diagnosed with EDMD, Sam's behavior and mental state changed significantly, but he remembers very little of that time; in retrospect, he realized he was likely traumatized by being diagnosed and quickly thereafter having surgery to implant a pacemaker.

> I remember being diagnosed and some things further, but apparently, there were a lot of things going on, a lot of things I was doing or saying, or not doing or not saying, from 10th to the end of 11th grade, that I don't actually remember. I can't really remember a whole lot between the ages of 15 and 16 . . . People usually have to tell me and fill in the details because doctors said sometimes if you can't cope with something your brain will shut the door and try to block it out.

Desmond reported repressing difficult memories of falling at school:

> It was hard because I would fall, or I would have lots of stuff like happen that just really got to me. I kinda tried to bury it down because thinking about it, it's not pleasant at all.

Confiding in others in depth about JOMD was not common in this sample. Participants had generally kept to themselves the mental and emotional struggles of living with their condition. Several participants had never talked in such detail about living with JOMD,

either because they assumed no one would understand, because they did not want to burden others, or because the experience was painful.

JOMD related anxiety. As evidence mounts that JOMD progresses, participants experience anxiety and dread. For some, panic attacks were reported. The fear of needing help and/or being helpless emerged in the interviews. The concern about falling and the inability to get up independently grew with progressive muscle weakness. Desmond was in high school when he could not get up from the floor without help. Each day he feared unexpected events, like fire drills, would render him dependent on others for help or display his disability to student and staff onlookers.

> I was just trying to get through high school . . . I have really bad anxiety right now, and that is where my anxiety took hold, there, is cause when I was walking, you know, any little breeze would make me fall. I would get so freaked out if there was a fire drill or something because everyone was crowded like cattle trying to get out.

At 24 years old, the fear of falling and not being able to get out of her car stopped Mae from leaving home by herself on a regular basis.

> I would like to go out by myself but I can't because . . . I am very afraid of falling . . . I can still drive but I can't . . . get out of my car sometimes by myself. I need to be picked up. So that stops me from going out and doing things with people.

The fears of inability to participate and rejection from society precipitated departure from higher education, foregoing of social opportunities, and avoidance or abdication of

employment. In other words, the cost of fear is isolation and social/occupational disengagement.

New social situations in unfamiliar environment were especially problematic and bothersome when the condition is invisible, while the limitations are real and debilitating. Lindsey describes her internal process as follows:

> When I have to go on-site to a new client or be in a new situation that's kind of outside my business, then literally my first internal thought is, "oh God are they going to be like 14 flights of stairs that I have to walk up in front of a new client?" or "Oh God are they going to know?" In new social situations—that's where my anxiety comes from.

JOMD related depression. Justifying JOMD to others is negatively anticipated and met with uneasiness. Though assistive devices and accommodations did help lower overall fears of needing help, they were approached with apprehension. The "justification to self" theme provides some insight into the reason for the uneasiness about accepting support.

Participants described sobering experiences that deflated once hopeful expectations about life. For example, falling triggered periods of reflection and emotional decompensation. Lindsey described her painful experience of falling in the grocery store parking lot, which is much different from what non-affected peers would experience in a similar situation.

> So, sometimes when I fall, I will go in my car and cry. I think the reason why is I have this big tough girl mentality and then all the sudden I get like dropped back

> down to reality of my shortcomings or what I am not able to do. And then I think about "why did I fall? Oh, my god is it getting worse?"

Loss triggered periods of sadness, self-deprecation, and social comparison. Lani described loss as a "bubble busting" exposing a harsh reality of personal incapability. JOMD was described as a condition that "mentally shattered" Sam, as he had to give up his career dreams upon being diagnosed. Repeated losses, compounded over time, impact mental health and interfere with social adjustment. Desmond described the mental toll it takes to be disappointed again and again because of living with JOMD, continually losing function.

> Why am I working so hard towards something when it is going to inevitably happen? It's like you try and try and persevere and you just hit brick wall after brick wall after brick wall and after a while it just takes a toll on you. I can't do it anymore.

The losses are compounded and vary in intensity. At the age of 29, Lani had endured losing much of her muscle function and independence, creating a low-grade persistent depression and core belief of worthlessness.

> I can't hide my tears. I cry a lot . . . there is always a little bit of depression deep down inside that I can't overcome and there are for sure days where I will be very down the whole day . . . there are days where I really tell myself, hey, you're not worth a thing. You can go and throw yourself somewhere and then it would be over. You wouldn't have to struggle with life because sometimes it can be really tough.

The degenerative nature felt defeating and incomprehensible, perpetuating existential angst as narrated by Mae below.

> I think the biggest thing is just wanting to live. There are times I feel—I get so depressed that I don't want to die or anything . . . what is the point of being here if I can't help myself and always have to rely on other people? So, I think that is the biggest emotional thing is just why are we made like this when there is no point?

Emerging adults are grappling with intense feelings of grief from JOMD. Low motivation made it difficult to re-enter higher education or the work force after departure.

JOMD related fatigue. Fatigue proved to be a frustrating aspect of living with JOMD for this sample, because it limited social interaction and engagement in the work force. Day to day, emerging adults with JOMD become aware of the energy certain activities, like walking up a set of stairs or going to a concert, require. As muscles deteriorate, participants compensated by being more selective in activities and social outings. Desmond expressed his internal process of weighing the value of going out with the energy it will cost him:

> If I know I am going to have fun, that the fun is going to outweigh how difficult it is, then I'll go. But if I . . . am not really that into it, then why am I going to . . . as negative as it sounds, why am I going to put forth all that effort because it physically takes a toll on me.

Jemma articulated how fatigue interfered with her ability to be physically active for two consecutive days:

> I save the amount of energy I do have, but let's say there is a little more physical activity that I would like to do today *and* tomorrow, I would have to choose between the two, because I know I am not going to be able to do something tomorrow if I exert a lot today. My muscles they are not going to have it. They are not going to be able to function even the way I can tomorrow if I try to do something today. So that is something I have recently had to learn to deal with.

Fatigue was consistent across participants, but varied in intensity. Around age 21, Jemma's symptoms became more pronounced as did her lack of energy. "I am always tired, I am always sleepy and stuff. That is another thing for motivation I just literally go to sleep and just like to lay in bed. That is where I am most comfortable." She attributed fatigue as one of the main contributors to her divorce; her husband liked to be socially active, but her fatigue impeded going out multiple times in a weekend.

The progression of JOMD is both a physical and an emotional challenge. In this sample's experiences, psychological aspects were rarely addressed in the treatment of JOMD. The main negative emotions that were described were repression, anxiety, depression and fatigue. Participants varied in degrees of physical impairment, which did not directly correlate to emotional health. Over time, negative emotions can lead to clinically significant symptoms and diagnosable mental health impairments. Of the eight adults interviewed, four had previously sought help from a mental health professional. One was in the process of attaining counseling services, but had not yet attended a session. For those who had previously been in treatment, only one found it helpful.

Theme 5: Fear of the Future

Fear of the future because of JOMD was reported by every participant, mainly due to uncertainty regarding the progressive and degenerative nature. Thoughts of the future elicited fears of severe disability and dependency. For those living with JOMD, the future is filled with impending physical decline that is uncertain in time and place. Certainty of future loss makes it a challenge to appreciate the abilities one has in the present, as Sam describes:

> It's not as physical as people think, even if you are in a wheelchair or able to walk, or whatever the condition may be of your mobility . . . because even if you are able to walk, you don't know if you will be able to walk later in life; and if you are in a wheelchair now, you don't know if you'll not be able to roll your own wheelchair later in life. There is no information and they don't know anything and can tell (me) anything. That doesn't mess with (me) physically, it messes with (me) mentally.

Emerging adults with JOMD are grappling with the fact that present ability is equal or superior to any future ability. Participants worried about future loss. Jemma articulated her fears of never-ending progression:

> The only thing is worrying, maybe, just about the future . . . I just wonder what life is going to be like for LG with the treatments or with me personally. I'm worried that things will get worse or . . . that I'll suddenly have a decline in energy or weakness and I don't know how . . . many times am I going to come off a plateau and it's going to get worse. How many of those do I have left?

Planning for the future is difficult when one is unsure how quickly muscle function will deteriorate or how much energy will be available to achieve personal and professional goals. To try to figure it out, participants measured losses since early adolescence and feared equal or worse progression in next decade or two. Lindsey described feeling anxious when she projects into the future, fearing what is to come:

> Well I guess I could say that scares me the most is when I don't feel on the day to day getting worse like you don't feel it's just something that happens over time, so when I stop and I look back what I used to do when I was 13 (years old) to now—that's a huge gap. You know and then you know you also ask yourself questions about like well if that happened in 14 years, what's going to happen in the next 14?

It can be difficult and depressing to speculate about the future. Denise explained how her worries have changed over time from peer acceptance to planning for her future with LGMD:

> I have different things to think about than when I was younger and it was mostly like peer stuff, but now it's . . . more-or-less like what is the rest of my life going to look like and how can I figure it out?

Jemma echoed a similar doubt in her circumstances and in her ability to cope with future progression of losing the ability to walk or drive without assistive devices:

> Am I going to be left somewhere, where I don't feel like walking, or (if) I have to stop driving or even get a hand assist vehicle. I worry about when and how I am going to deal with it mentally.

Participants demonstrated little or no hope that symptoms would improve or stay stable over time. Those who were mobile had images of themselves in wheelchairs which triggered anxious feelings. Those in wheelchairs projected difficulty breathing or being completely immobile. When projecting ahead in life, Desmond had thoughts and fears of total dependency:

> I try not to think about it (i.e., the future) because that would probably depress me because I don't want to know how bad . . . right now I am on a breathing machine at night and knowing . . . I could be bed ridden, ya know, and I hate to think about anything like that because I don't want that. So, I try not to think about it and just take it day by day.

Lani describes similar fears of being totally helpless by middle age:

> I was scared of the future, of how my life will go on, and I also asked myself if one day I will only be able to move my head or whatever. There came up so many pictures in my mind of what might come along.

It is as though emerging adults living with JOMD are unsure of their own prognosis.

For those who were not yet in a committed romantic relationship, participants assumed JOMD was a formidable condition that would deter most people from long-term partnership. Mae articulated her fear of being alone in the future, and not having family to depend on:

> In the future, I want to have a partner. I am a very big believer in love and having someone to be happy with. At least that makes life easier, having that partner, and I'm scared that I won't find that. So, I don't want to be alone when I

> am older because I'm not going to have my parents and my sister will have her own life. I don't want to be alone that way.

Desmond had similar fears of being alone, and seeing little value in himself and his ability to provide for a partner and/or child.

> I always go back to the MD that is affecting everything. If I were standing next to some guy, who would choose me over that guy? I mean . . . he is perfectly healthy . . . he can walk around . . . he can drive . . . what do I have to bring to the table? I always come up with little to nothing. Like I have got nothing to bring . . . if you stand me side by side with some guy, who is walking and stuff, I ask myself why would she pick me over him? Why would she put up with the wheelchair and having to transfer, helping me with a lot of stuff? Why would you choose that route?

The fears of being alone in the future were not just about feelings of isolation and/or rejection, but also of not being able to live independently and take care of one's own basic needs.

Finally, uncertainty regarding future ability makes additional developmental tasks, such as being a parent, arduous. Lindsey feared her condition jeopardizing her child's safety as he becomes more mobile as a toddler.

> What I'm really anxious about now is being able to pick up my kid off the floor when he starts to crawl. That's the kind of anxieties as a new mom you know like how am I going to do this? How am I going to run after my kid when he starts running towards the street?

Simple tasks such as picking up a small child are impossible for some individuals with JOMD. It is hard to plan how to circumvent physical obstacles independently, without feeling overwhelmed by negative feelings.

The future is a scary and uncertain place for emerging adults with JOMD. When participants were asked how they believed JOMD would impact their future, the anxiety and frustration about living with JOMD was unmistakable. The challenges of the past and present were perceived as more manageable than the seemingly formidable challenges that can be expected of a future with JOMD.

Summary of Chapter III

This study uncovered five themes that captured the experiences of emerging adults living with JOMD. The first theme, "JOMD expands into consciousness with continued loss," featured how changes in physicality require more consideration about the illness; JOMD begins as an afterthought and by emerging adulthood had a bigger influence on the individuals' lives. Secondly, participants shared the stress of experiencing increased visibility of JOMD, both to themselves and others. Discernibility lead to theme three—self-other justification of JOMD—or how affected individuals make meaning of bodily changes. "Evolution of disease, evolution of negative emotion" was the fourth theme to emerge, which included four sub-themes composing different emotional challenges because of living with JOMD: (a) suppression, (b) anxiety, (c) depression, and (d) fatigue. Some participants perceived themselves to be more emotionally than physically affected by the illness. The last theme, "fear of the future," comprised the shared experience of fear as these young adults explored what is to come.

The uncertainty of progression and ambiguity of prognosis lead to mostly negative anticipation from present to future.

In Chapter 4, the researcher describes the relevance of these findings to the field of Counselor Education and Supervision. Results are compared with existing literature on the topic of emerging adulthood and pediatric onset slowly-progressive neuromuscular disease. Implications for counselor educators, supervisors, and practitioners are offered, along with limitations and areas for future research.

CHAPTER IV

DISCUSSION

The purpose of this study was to understand how emerging adults perceive and describe their experiences of living with JOMD. The phenomenological methodology provided an opportunity to gather detailed, first-hand, accounts of the experiences of eight individuals in their 20s living with either LGMD or EDMD. The researcher aspired to gain insight into the psychological aspects and consider how counselors might be helpful in short- and long-term care. As presented in chapter 3, the experiences of emerging adults living with JOMD are described in detail in five common themes shared across participants: (a) JOMD expands into consciousness with continued loss, (b) the stress of experiencing increased visibility, (c) self-other justification of JOMD, (d) evolution of disease, evolution of negative emotion, and (e) fear of the future. These themes are reflective of personal, yet shared, experiences that are rarely addressed and highly relevant to mental health counselors.

Emerging adults describe living with JOMD to be like "hitting a brick wall" repeatedly, where what one thought to be able to do routinely, or at least periodically, collides with reality. The experience is filled with untimely losses that trigger a justification process, or explanation of weakness to oneself and others. Perceptions of other people's negative judgments were expressed, and corresponded to the stress of increased visibility. Over time, the losses compound and one realizes more and more what it means to live with JOMD. Participants described times when suppressing thoughts about their condition helped in remaining optimistic about future health.

However, the discovery of new limitations increased awareness of progression and was psychologically distressing. Common emotions reported were suppression, anxiety, depression, and fatigue. The mental and physical effort to live "normally" increased over time. Finally, the emerging adults in this study described fears about how JOMD will impact their future (e.g., what losses are yet to come and how will I cope with it?). Treatment from informed, person-centered mental health counselors, especially during times of instability, such as a shortly after diagnosis, noticeable physical progression, change in residence, departing work/higher education, or graduating from college could assist in adjusting to the illness. Learning ways to communicate to others about the condition would also be useful.

The purpose of this chapter is to discuss the meaning of the data and address implications and limitations. The results are then compared and contrasted with the existing literature in the domains of grief, emerging adulthood, juvenile onset chronic illness, and MD. Next, how the results of this study contribute to the literature are described, followed by implications of the findings for Counselor Education and Supervision. Finally, limitations of this study and recommendations for future research are explored.

The Essence of Emerging Adults With JOMD

The intent of conducting this qualitative research study was to understand and describe shared aspects of emerging adults living with JOMD. Little research was found in the literature on the mental aspects of living with non-congenital forms of MD from the perspective of emerging adults. What does exist focuses only on LGMD2a (Aho et

al., 2015, 2016, 2017). Thus, this study was conducted to investigate the illness experience of JOMD, a condition that progresses significantly between the onset of symptoms (around age nine) and age 29. The research findings from the eight participants interviewed provide rich and meaningful data about what it is like to live with JOMD.

The following section summarizes and discusses the data as it relates to the existing literature, guided by the emergent themes of the study. Similarities and differences of the results are explored. Though the two forms of JOMD represented here are clearly distinct in genetic and medical terms, the themes reflect the shared experience.

JOMD Expands Into Consciousness With Continued Loss

Within the first theme, "JOMD expands into consciousness with continued loss," it became apparent that to live with JOMD is to continually adjust to new and evolving losses or limitations, which triggers a cycle that, over time, impacts consciousness and mental health. A loss/limitation experience moves JOMD into consciousness, or into newfound awareness of how the condition impacts life, which then elicits a justification response. For example, when Desmond was a senior in high school, he accidently tripped on an area rug and tore a ligament in his knee, one injury among many he vividly remembers. He then became much more aware of rugs and learned to be cautious to avoid repeating the experience—begetting growth into consciousness. His emotional state decompensated after he was unable to attend a school dance, feeling angry, upset and inadequate, byproducts of his justification process. He described what he was

thinking and feeling as he processed how disabled he became from one fall, and how the injury exaggerated his muscle weakness in the following quote:

> I finally started talking to a girl and got her to go to a dance with me, and this just stupid disease is screwing it all up. Why did this have to happen? Why? It was always more so in the short term, like why did . . . I have to trip on the rug? Why did it have to happen at this moment? And then, if I would sit there and think about it I would go . . . Why can't I just be normal? Why can't I get up and walk? My brother can walk, why can't I? . . . I remember I was just so devastated.

Because JOMD is particularly degenerative during late adolescence and emerging adulthood, the process is repeated. A loss, such as a fall or trouble climbing stairs, signifies an aspect of disease progression, while the growth into consciousness through the justification process is a psychological response. A simple depiction of the process can be viewed in Figure 3. While the degree of the impact of the loss varied, the volume of losses increased significantly between adolescence and emerging adulthood. To put it another way, losses or limitations are like force on a gas pedal that propels a cycle of thinking about what it means to live with the illness. When progression is most noticeable, like travelling to a new place, the cycle accelerates as the person realizes that symptoms are impacting life. This can cause them to justify why it is happening and what it means. The metaphor of "hitting a brick wall again and again" is another way of describing the experience of realization, grief and change as losses perpetuate over time.

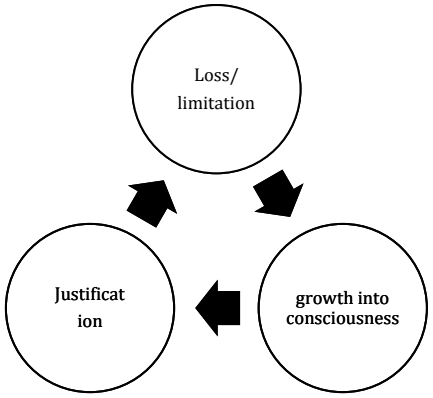

Figure 3. Chronic loss in JOMD

Previous research studies have described the endless adjustments required to live with progressive neuromuscular disease (Aho et al., 2015; Krause-Bachand & Koopman, 2008), but none articulate the culmination or intensification of losses adding up over time, leading to changes in consciousness. The gradual nature of JOMD distinguishes it from other juvenile onset conditions and it impacts adjustment, because one's health is continually changing and worsening. Verhoof et al. (2014) demonstrated that young adults with a positive or stable course of illness reported more acceptance and less helplessness than those with a variable or degenerative disease. Results from this research study help us understand how the course of this illness impacts a person physically and mentally, and helps us understand the unique challenges of progressive disability.

Loss is a crisis commonly experienced by those with chronic illnesses, and triggers a process of grieving for the reduction of functioning (Livneh & Antonak, 2005). Living with a disability evokes loss of status, social support, functioning, and control, all losses described by participants (Smart, 2009). The degenerative nature of MD makes

loss unavoidable, increasing the likelihood of chronic sorrow—the periods of sadness or grief, alternating with times of stability and happiness, as one realizes "what is" versus "what should have been" (Nätterlund et al., 2001). Results of this research show losses big and small: The inability to pick up dropped grocery store items, using the toilet independently; the dream of being a fireman; going shopping alone; picking up an infant child. It is estimated that the rate of chronic sorrow among persons with Parkinson's disease is 50%, and those with multiple sclerosis is 83% (Nätterlund et al., 2001). Yet, this concept is very rarely addressed in the MD or pediatric chronic illness literature.

Results of this theme align with current theories on loss and bereavement, suggesting that growing up with JOMD is as much or more of a grieving process than one of psychosocial adjustment. Through the Dual-Process Model lens, declines in physical functioning trigger a dominant loss-orientation, which consists of JOMD growing in consciousness and the justification process that follows. The recurring loss captures the negative emotions associated with bereavement, and results also extend beyond the model to expose the other normative experiences of living with JOMD, such as anxiety, depression, and fear of the future. The restoration-orientation depicts the stressors that are a consequence of loss of functioning, such as avoidance of activities that are associated with increased visibility. Denial and suppression of feelings related to JOMD are other coping processes utilized in restoration.

While the recurring losses that happen because of living with JOMD mirror the experience of bereavement, the continuing obstacle of habituating to evolving deteriorations are unique to the degenerative course of the illness. The negative emotions

that arise over time, in and of themselves, are not necessarily detrimental to adolescents and young adults with JOMD because it is through this suffering that growth occurs. However, some participants describe stagnation in loss during periods of higher stress, such as when being diagnosed, when increased visibility occurs, and when an assistive device is required. Those with a more aggressive course and faster muscle breakdown were more likely to report intense grief and loss. Similarly, those with little physical impairment were minimally engaged with the loss and more invested in reorientation.

An undertone of "meaning making" and "reconstruction in the face of loss" is echoed in the results. Progressive losses propel the need for persons to build new meaning structures about chronic illness and disability and adjust to changing circumstances. Through the justification process participants enter an appraisal mode that is captured in the sense making and benefit finding activities described in the Meaning Reconstruction Model (Gillies & Neimeyer, 2006). The psychosocial changes that occur from the justification process naturally fit into the constructivist theory. The JOMD related depression sub-theme catches a glimpse of the existential questioning that occurs during the reconstruction process. It is currently unclear how those with JOMD make meaning out of their experiences. As finding benefits to a tragic experience generally does not arrive until months or years later (Gillies & Neimeyer, 2006), perhaps finding benefits in JOMD and integrating the condition into one's identity occur later in life, such as the 3rd–4th decades of life.

The research done by Thannhauser (2014) among the pediatric multiple sclerosis population was the first to emphasize the experience of grief in understanding children

and adolescents with chronic illness. The Grief-Peer Dynamics theory was developed to describe the notion that adjustment is continuously influenced by loss and peer relationships (Thannhauser, 2014), and results of this study are among the first to strengthen that notion with respect to the emerging adult MD population.

As one becomes more conscious of the muscle weakening, the cognitive and physical effort to adapt to the environment increases. In other words, one has to think more about how to get around obstacles to live as "normally" as possible. If there is a social event, a person with JOMD will consider details invisible to the non-affected peer, such as if there are stairs at the entry, a first floor bathroom, and the height of the toilet seat. Over time, these obstacles mentally build. Some participants demonstrated cognitive fatigue, or mental exhaustion from working so hard to manage day to day life. The stress-diathesis model suggests that affected individuals living with JOMD are therefore at increased risk for a predisposed mental or emotional disorder to emerge (Burke & Elliott, 1999). This research is among the first of its kind to explore the inner lives of emerging adults with JOMD without using quantitative instruments that cannot capture unique aspects of this particular condition. It does support the notion that in these cases learned helplessness contributes more to psychological functioning than disease severity (Evers et al., 2001; Nieto et al., 2012), further supporting a neglected chasm in traditional medical treatment.

Vash (1998) wrote about the role integration of an individual's disability with consciousness development. An interesting and relevant point here is that conditions, that leave the lifespan unaltered but deeply threaten one's ability to be independent,

able-bodied, and free of pain, lack, and loss (e.g., JOMD), accelerate psychological development. Distant secondary experiences that also serve to catalyze growth of consciousness are meditation, psychotherapy, intense athletic or artistic achievement, psychedelics (in rare cases), yoga, and other focused practices on mind-body connection (Vash, 1998). From this perspective, a life-threatening chronic illness gives someone a major advantage or head start on considering what it means to be normal and live a meaningful life. Vash (1998) explained that catastrophe, dysfunction, and suffering motivate humans to evolve. The essence of this idea is present underneath the experience of "hitting a brick wall" and "rebuilding" oneself time and time again. Why rebuild? Why keep trying when the "inevitable" physical declines will happen anyway? With a body that slowly declines, it is no wonder why those with JOMD attend to cerebral matters—enhancing psychological demands and opportunities to find new meaning in a difficult situation.

To live with JOMD is to experience repeated losses that require both physical adaptation and cognitive restructuring. Results of this study describe how JOMD presented itself to consciousness in minimal ways in childhood, and how it changed over time to become a bigger part of everyday life. This sets this research apart from much of the preexisting research that gathers a snapshot of various concepts without a deeper contextual understanding, which explains why the notion of loss and grief went undetected in the existing JOMD literature. Gaining a more accurate understanding of the phenomenon opens treatment options that were not previously available. Neglecting the idea of grief in understanding of MD, and assuming adjustment unfolds similarly to

other chronic illnesses is damaging and stigmatizing to the experience of the repeated losses that occur over time in the case of patients with JOMD. Results here normalize grief and suggest future research is needed. The suffering that comes with JOMD could also be reframed as an advantage, or an opportunity to take on new roles, have greater self-awareness and gratitude, be more empathic, and develop wisdom (Gillies & Neimeyer, 2006). Finally, processing these losses requires thought and energy, which is occasionally exhausting for some participants, as they simultaneously carry on life as usual, attending college, work, and having children.

The Stress of Experiencing Increased Visibility

In the second theme, the stress of experiencing increased visibility, the struggles of having a disease that begins as nearly indiscernible and slowly becomes visible, was brought into focus. Several studies among mixed samples of adults with MD briefly describe the self-deprecation and shame that corresponded with increasing visibility of symptoms (Bakker et al., 2017; Boström & Ahlström, 2004; Nätterlund et al., 2001), though none have examined the progression and subsequent stress associated with symptoms. The discomfort of the illness being detected may be intensified because symptoms are getting worse during adolescence and young adulthood, times in the lifespan where one particularly wants to belong and fit in with peers. These results expand upon the transition from the invisible to visible nature of JOMD and the experience of symptoms becoming more obvious to self and others by one's 20s. See Figure 4, Desmond's Progressive Experience, for further clarification on how progression, visibility, and stress interact.

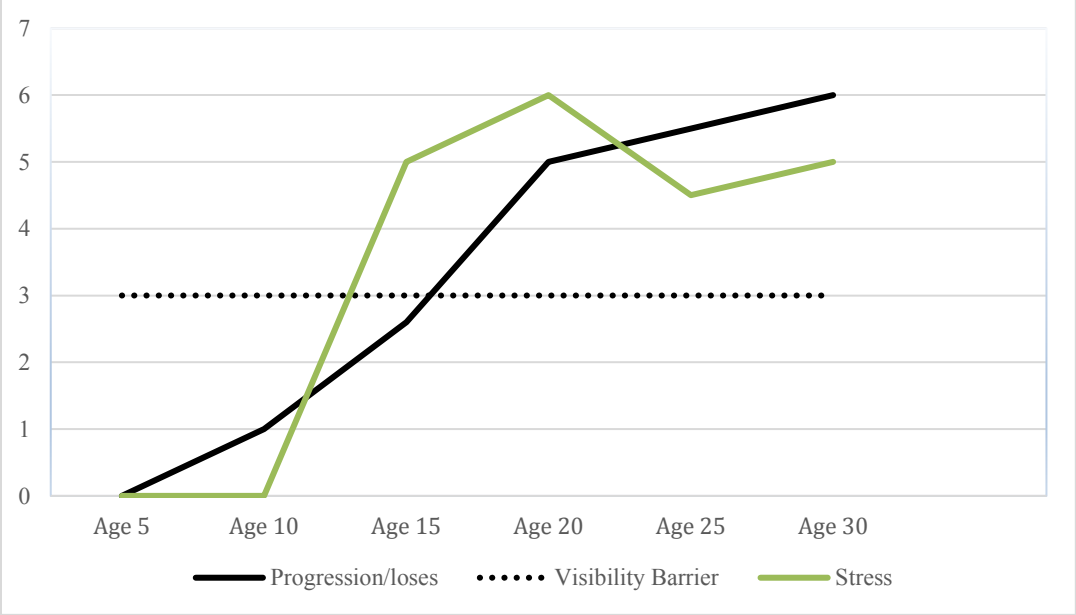

Figure 4. Desmond's Experience of Progression

The time where the progression/losses and visibility to others intersect, as well as the months/years leading up to and following it, are periods of high stress. For Desmond, his stress went up significantly between the ages of 10 and 20, as he went from mild muscle weakness to requiring a power wheelchair—and all the subtle stages in-between. Stress decreased after he received accommodations and left work and school, but new stressors, such as lacking purpose and meaning and growing hopelessness he will find a romantic partner, account for the rise from ages 25 to 30. The age at which point progression/losses intersects with the visibility barrier varied between participants, but a general rise in stress preceded and followed the junction of variables.

Van der Beek et al. (2013) studied the influence of stigma on quality of life among 235 patients with neuromuscular disease and found that shame and fears of discrimination were more common and detrimental to quality of life than actual

exclusion. The degree of conspicuousness of a disability is one dimension of stigma that is associated with prejudice (Smart, 2009). Disability researchers have incorporated this finding into ecological models of disability, which assist counselors in determining important factors for psychosocial adjustment (Vash & Crewe, 2004). One sub-factor of the nature of disability is its visibility. When a disability is obvious, such as requiring a wheelchair for mobility, individuals report discrimination, devaluation, or being ignored (Marini, 2012b). Those with invisible conditions, such as mild muscle weakness, are misjudged as lazy, unmotivated, and even deceptive for not participating in certain activities, like employment (Smart, 2009). Therefore, people with JOMD are under tremendous social pressure to "pass" as someone without limitations. Initially not apparent to others, it is only the accommodations that are eventually required (e.g., wheelchair; cane; pacemaker) that inform others of the limitation.

Discrimination against individuals with chronic illness lowers the likelihood of achieving adult milestones (Smart, 2009). Pinquart (2014) found that achievements like attaining higher education, employment, leaving home, and getting married have significant moderating effects on visibility of the illness. Results of this study are consistent with Pinquart's findings, in that those who completed college, had a stable, significant romantic relationship, and lived independently, had a more positive perspective on life with MD.

Furthermore, data from this study show that muscle weakness initially brought negative peer attention and bullying encounters in school. Several participants reported some of their most emotionally distressing times being between 6th and 12th grade, at

times when the condition was becoming more obvious to others. No research, to the author's knowledge, exists about peer exclusion specifically related to JOMD. Findings from this study suggest that middle and high school students with mild neuromuscular disorders are at risk of peer discrimination and social isolation.

Insight into the effects of the transition from the invisible to visible nature of the disease symptoms is another unique contribution to the literature in understanding emerging adults with neuromuscular disease. Both stigma towards oneself as a person with MD and perceived stigma from others are stressful to this population growing up with a debilitating condition. Noticing symptoms of JOMD as a child, adolescent, and young adult is much different from being diagnosed in mid-life, where one has a more formulated identity and time to achieve stability in a career and/or relationship. The progressive loss of physical functions adds significant stress in planning for the future, and obstructs a once limitless worldview shared by many emerging adults (Arnett, 2015). The degree to which these youths succeed in navigating the perceived stigma has major consequences for the accomplishment of developmental milestones, such as completing an education, living independently, and establishing a long-term romantic partner relationship.

Self-Other Justification of JOMD

In the third theme, "self-other justification," a private cognitive process was discovered as part of the illness experience. This is an important finding, because early recognition of negative illness appraisal might be a key to the identification of patients at risk for psychological distress and reduced quality of life that is reported among the adult

MD population (Padua et al., 2009; Verhoof et al., 2014). There is a connection between how participants "talked themselves through" a stressful scenario when JOMD was brought into intense focus (e.g., public fall) and emotional well-being. It would be beneficial to increase affected persons' and their families' understanding of progressive disability in order to separate symptoms of the disease from perceptions of lack of effort, laziness, or carelessness. See Figure 5 for a general visual interpretation of how the justification process unfolded.

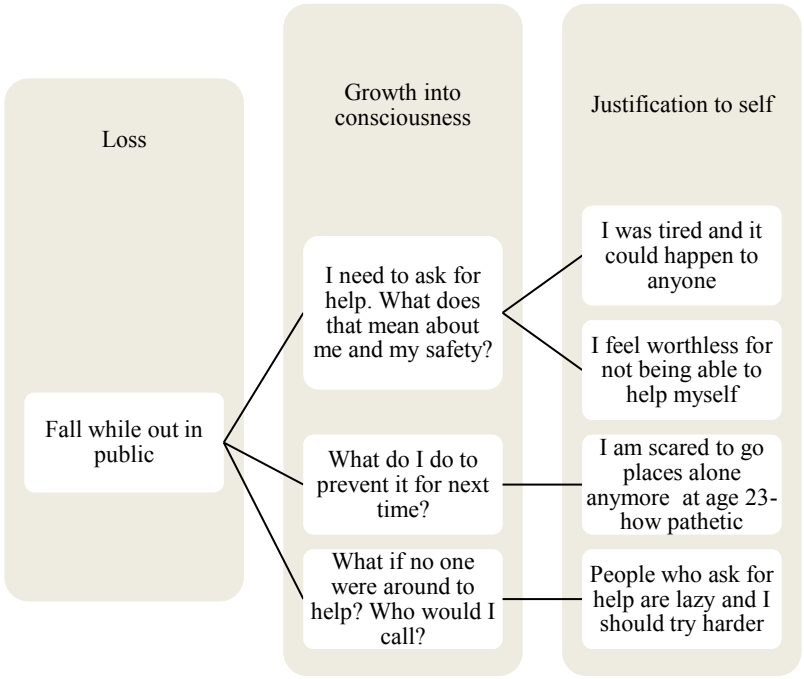

Figure 5. Loss, growth and justification process

It was interesting that several participants describe accepting help or utilizing an assistance as "giving up" on themselves and/or their dream of not being seriously disabled. These results could be compared to the "fighting spirit" promoted among cancer patients and assumed by some adults with MD (Ahlstrom & Sjödén, 1994).

Assuming a win-lose mentality sets a person with MD up for failure and disappointment. Disease progression is unavoidable, and a hallmark feature of MD, yet little attention is given to explaining to the patient exactly what it means and how it manifests itself in real life. Affected young persons are left to themselves to make sense of the experience. In this study, those who were not using a power wheelchair had a negative, or, in some cases, catastrophic expectation of the experience. Similar findings were published in Aho et al. (2018). Results of this study suggest that misinformation and lack of awareness about MD impacts how one perceives and justifies it.

Lack of awareness among the public created barriers to communication for the participants in this study. Participants reported negative experiences when telling others about the condition, and were met with skepticism about requests for accommodations or given unwanted and misguided advice when explaining a limitation. Invalidating experiences and fears of rejection limited social engagement in every participant.

Barriers to communication also existed within families. It was up to the affected young adult to educate others about the illness, thus the patient never reached a level of "feeling understood" regarding the experience of living with JOMD. This study seeks to minimize that gap. Justifying symptoms to others is less of an issue with sufferers of the birth/early onset types of MD, as their disability is more obvious from a young age. This sample expressed great difficulty explaining their condition to others.

Evolution of Disease, Evolution of Negative Emotion

Emotions such as anger, irritation, depression, anxiety, and panic have been noted among adults with MD (Boström & Ahlström, 2004; Nätterlund et al., 2001), and the

results of this study expand upon the literature regarding the mental and emotional impact of MD. Possible explanations for contradicting results in the pre-existing literature include (a) the wide range of ages in the sample, taking away from the distress of the developmental period; (b) the instruments used to gather data on symptoms such as depression and anxiety; and (c) a possible unwillingness to disclose such personal details; or (d) the perception that talking about the emotional impact would cast a negative social light. A group of men with Duchenne MD aged 15–33 reported wanting to protect parents from their distress, or feeling as though such topics were "off-limits" (Abbott & Carpenter, 2015). It appears that the same sentiment was experienced within some of the participants' families in the present study. Such an omission or underreporting downplays the issues, perpetuates the lack of sufficient services and masks treatable problems.

The sub-themes are a collection of shared narratives from emerging adults themselves living with JOMD about unsettling emotional aspects. As emerging adults reflect on their experience of living with JOMD, it was a theme that emotional responses and issues negatively changed since the time of symptoms onset.

JOMD related denial. The models of loss described in the literature review also have relevance for the emotional experiences described within this theme. The Dual Process Model explains that repression/denial experienced by emerging adults with JOMD is a means of coping with the stress of unavoidable loss, and is a necessary aspect of the grieving experience (Stroebe & Schut, 1999). Accepting loss as an essential component of JOMD, it would make sense, from a Dual Process Model framework, that

coping would also involve taking time to separate oneself from the loss and becoming engaged in other, more rewarding or comforting, endeavors. Both avoidance and immersion in loss are strategies for coming to terms with this life event, and it is when the oscillation between these two processes ceases that mental health counseling may be necessary. Too much suppression of negative emotion, or sustained effort to carry on as though nothing has changed, has adverse health effects such as high blood pressure and heart rate abnormalities (Stroebe & Schut, 1999). On the other hand, a chronic loss orientation could signify complicated or prolonged grief, creating its own set of problems (Stroebe & Schut, 1999). Traditionally, prolonged grief disorder is defined as a persistently elevated set of grief symptoms among bereaved individuals that include separation distress, withdrawal from relationships, sense of meaninglessness, and difficultly functioning day to day (Simon et al., 2007). Other negative outcomes of prolonged grief disorder include symptoms of anxiety and depression, heart problems, high blood pressure, changes in eating and smoking habits (Prigerson et al., 1997). No research exists examining prolonged grief within the MD population.

Denial is one of the most frequently experienced psychosocial reactions to chronic illness (Feldman, 2011). The Stage Model of Adaptation to Disability describes 'defensive retreat' as part of a gradual assimilation process of an acquired disability (Smart, 2009). Denial has been found to center around the presence, implications, and permanence of the disability (Smart, 2009). The results of this study depict wishful thinking, especially following the first years after diagnosis, that symptoms would not get worse and that the illness would be minimally intrusive. Denial serves as protection from

threatening emotions, and can prevent "emotional flooding" or feeling overwhelmed by information about the diagnosis. Symptoms of JOMD were typically experienced in late childhood/early adolescence, a time when body image is most important and self-esteem is particularly vulnerable (Livneh & Antonak, 2007). Being "different" is experienced as devastating and disruptive to positive adjustment. Thus, it is no wonder why distancing oneself from stressful, unresolvable, thoughts and feelings is a form of emotion-focused coping previously described among the adult MD population (Ahlström & Wenneberg, 2002) and used among participants in this sample. Nätterlund et al. (2001) previously reported a group of adult myotonic MD patients who denied their condition or avoided learning and talking more about it for fear that it would impact their future. Explanations for avoidance in this study are fear of burdening others, perceived helplessness, expected rejection/humiliation, or perceived inability to cope with emotions.

JOMD related anxiety. Anxiety is one of the most common disorders reported among the young adult population, and those living with progressive muscle disease are no exception (Ferro et al., 2017). In the interviews, topics such as surgical implantation of a pacemaker, falling (especially during middle and high school), and being negatively judged were described as anxiety provoking. Circumstances in which the disability became noticeable to others who did not know about the diagnosis also generated feelings of anxiety. Several participants had previously sought counseling services due to symptoms of anxiety/panic attacks, and more were interested in acquiring them. For these participants, anxiety was also associated with asking for help or needing an assistive device.

Psychological disorders, like depression and anxiety, not only have been shown to worsen quality of life, but also exacerbate disease progression and contribute to autonomic dysregulation (Sabharwal, 2014). Recommended therapeutic remedies typically include medications to regulate physiology, with little attention paid to the power of naturally regulating the stress response through therapy. It is particularly important for the JOMD population to understand the stress response and its effects on the body. No research to date has examined the impact of counseling and MD symptom progression.

Success in the areas of love and work typically mark the end of emerging adulthood, and it is precisely these areas that elicited feelings of uncertainty of how JOMD would impact successful completion of these steps into adulthood. Mitigating anxiety and helping affected individuals cope with aspects of JOMD could improve the rate of higher education completion, establishing employment, and getting married, milestones that correlate with quality of life and emotional health (Maurice-Stam et al., 2007).

JOMD related depression. A natural response to repeated, untimely physical losses, especially in a stressful developmental period where most people reach all-time highs of fitness, triggered periods of intense sadness, inadequacy, and existential questioning. Previous literature indicates that individuals with childhood onset chronic disease are prone to experiencing a depressive episode, or exhibiting more severe symptoms that greatly affect everyday life, compared to healthy peers (Kokkonen et al., 2001; Secinti et al., 2017). Compared to their Duchenne MD counterparts, who most

frequently experience depression between 8 and 11 years old (Elsenbruch et al., 2013), those with JOMD are encountering this distress later in life—between adolescence and emerging adulthood.

In this study, symptoms of depression came and went with participants at various times, but were poignant during compounding loss and environmental changes. In slowly progressive neuromuscular conditions, mental health problems had more impact on health-related quality of life than physical symptoms (Dany et al., 2017; Perrin et al., 2007). Findings from this study suggest that periods of depression are a normal part of the MD experience, yet are rarely addressed or treated.

Half of participants spontaneously spoke of a history of suicidal ideation during periods of depression. It is unknown if those participants had also made a plan or past attempts to commit suicide, as it is unknown if those who did not bring it up also had thoughts of suicide. Perhaps such questioning is a symptom of depression, or an aspect in grief, or both, among this population. Being diagnosed with JOMD is to get a life sentence of physical deterioration, subsequently throwing assumptions about oneself, the definition of success and what brings joy, into question. Collectively, these preliminary results suggest suicidal ideation is prevalent among emerging adults with JOMD. More research needs to be done in this area to get a more accurate depiction of suicidality among those living with JOMD between the ages of 12–29.

JOMD related fatigue. The study findings provide new information regarding how fatigue is experienced. Fatigue is defined as an overwhelming and sustained sense of exhaustion that debilitates functioning and ability to carry out daily activities, above

and beyond muscle weakness because of JOMD (Matura, Malone, Jaime-Lara, & Riegel, 2018). Underlying biological factors causing fatigue for individuals with chronic diseases such as multiple sclerosis or cancer include inflammation, autonomic nervous system activation, and hypothalamic-pituitary-adrenal axis dysregulation (Matura at al., 2018). Consistent with previous studies of persons with facioscapulohumeral MD (Smith, McMullen, Jensen, Carter, & Molton, 2014), oculopharyngeal MD (van der Sluijs, 2016), and LGMD (Perrin et al., 2007), fatigue is a common symptom of MD that negatively impacts quality of life. Tiredness has been shown to increase thoughts of helplessness and decrease acceptance of disease (Verhoof et al., 2014). Results of this study underscore that exhaustion, both physical and mental, limits occupational and social participation—two milestones of profound importance in emerging adulthood. One possible explanation for fatigue includes recent over-activity (causing inflammation), which was likely to occur after diagnosis, but before the use of a walking aid or wheelchair. Another possible explanation is that the mental efforts involved in navigating the physical world with reduced muscle functioning may subconsciously activate the autonomic nervous system, especially as the condition becomes more stressful and difficult to manage without an assistive device. Negative effects of fatigue on the daily lives of emerging adults with JOMD include guilt and disappointment for cancelling social outings, dropping out of college, and even divorce because of lack of social activity. Fatigue reinforced negative perceptions of the self as lazy, unmotivated, and incapable of employment. Inability to participate socially can have grave effects, as friendship predicts both immediate and later resiliency (van Harmelen et al., 2017).

Fear of Future Progression

In the final theme, "ear of the future," participants describe anxiety and uncertainty when considering how life will change because of JOMD. People with chronic progressive conditions have reported fears of becoming disabled, and such fears are an important source of stress in these young people's lives (Kwakkenbos et al., 2014). Fears emerged in this study about future progression and not having needed physical support and medical services, are consistent with the findings of one other research study of emerging adults with LGMD2a, where uncertainty about future health was perceived as a negative consequence of living with the disease (Aho et al., 2015).

Emerging adulthood is typically a time in life when hopefulness for the future is at an all-time high (Arnett, 2004). In a national survey, 96% of emerging adults agreed with the statement, "I am very sure that, someday, I will get to where I want to be in life" (Arnett, 2004, p. 16). Comparatively, those with JOMD reluctantly and fearfully face the future. When prompted with the question: "How do you believe your condition will impact your future?" participants hesitated to speculate and disclosed deep fears of physical loss, reduced lung capacity, and early death, when looking at the road ahead. Knowing that progression is likely, but not when, or how severely disabled they would become, was a source of uncertainly uniquely experienced by individuals with JOMD. The ability to leave the family of origin, but not yet be in a committed relationship, is what prompts the "Age of Possibilities," another core feature of emerging adulthood (Arnett, 2004). The benefit of separation from parents and siblings, without yet having new commitments and obligations, is to experience an environment free from impositions

of family problems (Arnett, 2004). For those who had a difficult childhood, it is especially important to depart from the family home to "straighten the parts of themselves that have become twisted" (Arnett, 2004, p. 16). In the results of this study, all participants lived with either (a) family member(s) or a significant other, who assisted in caretaking responsibilities. There is an inherent tug of war between needing physical help, while striving for independence, that makes considering the future different and emotionally painful for the population under investigation. The imposed restrictions of having this illness complicate the path to adulthood.

The combination of "evolution of negative emotion" and "fear of the future" during adolescence and emerging adulthood is fertile ground for developing hopelessness, which has been identified as one of the most important risk factors of suicide ideation and attempts (Klonsky, May & Saffer, 2016). The concept of hopelessness was generated from the theory of learned helplessness and is defined as negative expectations about the future that are internal (i.e., caused by me not someone else), stable (i.e., will not change), and global (i.e., will impact many situations) in nature (Beck, Brown, Berchick, Stewart, & Steer, 1990). Negative life events and a negative cognitive style put people at greater risk for developing hopelessness depression (Zhou & Chen, 2017). Clearly, people with JOMD are more likely than most to experience negative life events and, to date, the complete lack of treatment to stop or reverse muscle damage imposes a significant challenge to developing hope. While neither positive nor negative life events alone can cause hopelessness depression, they are "occasion setters" in a chain reaction of developing depression, and can highly contribute to the

development of hopelessness (Zhou & Chen, 2017). The justification process highlights one's thoughts about the illness. Implications for this finding in counseling are discussed.

Contributions of Emerging Adults With JOMD to Counselor Education

A review of the literature illustrated a gap in understanding the experiences of emerging adults who live with a slowly progressing neuromuscular disease, particularly the cognitive and emotional responses that occur alongside physical progression. The degenerative, non-curable, invisible to visible, nature of JOMD distinguish it from other, more commonly researched juvenile onset illnesses such as diabetes, cancer or epilepsy. As a group, emerging adults with chronic disease are markedly underrepresented in the counseling research and few recommendations exist to address the mental health of this population. The results of this study uncover essential aspects of living with JOMD that provide a unique and necessary contribution to the counselor education literature.

Contributions to the Literature on Human Development

First, this study raises awareness and understanding of JOMD, an especially pernicious condition in which the severity of impairments and limitations during adolescence and emerging adulthood marks the patients as distinct from their chronic illness and age-related peers. Well-intentioned counselors might believe that the services required by young adults with different childhood onset chronic conditions are similar, regardless of the diagnosis. Although the basic foundational skills required for the counseling diverse individuals are similar, the nuances that lead to assisting these young adults in adjusting to their illness are important and can be a determinant in achieving

successful developmental outcomes. Results of this study suggest that the JOMD population is underserved, both because mental health is rarely addressed in the treatment of MD, and because the body of research regarding the need for counseling services is offering mixed results. This study offers a first attempt to increase the knowledge and awareness about this overlooked group and advocate for more comprehensive care for the individuals involved.

Another contribution of this study is that it describes differences between typical emerging adults and those with JOMD. One of the most central components of emerging adulthood is "The Age of Identity Explorations," during which emerging adults will experiment with various possibilities that determine preferences, especially in the areas of career and romantic relationships (Arnett, 2004). The process of identity formation intensifies during this developmental period. Living with JOMD inhibited opportunities to try out different jobs or college majors, and thus the ability to learn more about oneself. Some participants described their condition getting worse after high school, and experiencing unmanageable limitations that caused a premature departure from college. A change in residency highlighted progression and loss when everyday necessities, such as the toilet of shower, became new obstacles to navigate. Whereas the typical emerging adult may ask themselves, "What kind of work am I good at? What kind of work would be satisfying for the long term?" (Arnett, 2004, p. 9) those with JOMD are asking questions of "What kind of work is realistic for me in the long-term? How will I be able to do this job if/when my condition gets worse? Will I have the energy?" It is a privilege of most emerging adults to entertain endless possibilities without any restrictions. The

results of this study describe that, for emerging adults with JOMD, it is difficult to explore one's identity without frequent interruptions, of either mental or physical nature, that limit possibilities. This is consistent with earlier studies suggesting that young adults who have attained intimacy and identity formation before the onset of symptoms may cope better with the diagnosis than those who have not (Hauser-Cram et al., 2009).

The losses encountered during emerging adulthood by those with JOMD make the already notably unstable period of the lifespan even more intense (Arnett, 2004). The focus on having a well thought out plan, mapping the path to independence, is difficult when tremendous energy needs to be directed at thinking through a trip to a friend's home, navigating stairs and bathrooms, and getting up from a chair. While most 18- to 24-year olds do not know where they will be living from one year to the next (Arnett, 2004), those with JOMD reported fewer options for residential freedom due to mobility restrictions and possible need for caregiving assistance from family. Reasons for instability for those with JOMD may outwardly seem typical, but may be due to subtle progression of the disease and uncertainty about how to troubleshoot the resulting restrictions. For example, dropping out of college and moving home is not an unusual experience, but may, for those with JOMD, be due to difficulty getting up from a desk and fear of falling and needing help. How does one learn to become self-sufficient while activities of daily living require ever more thought and effort than years prior? In the United States, becoming an adult means accepting responsibility for oneself, making independent decisions, and becoming financially independent (Arnett, 2004). As results show, living with JOMD requires help. The gradual progression of the disease and

subsequent losses restrict independence: A devastating realization to arrive at for individuals living with JOMD that can deter motivation to revise plans and continue moving forward.

Contributions to Psychosocial Adaption to Pediatric Onset Chronic Illness

The experience of living with JOMD found in the results of this study is closely mirrors the Grief-Peer Dynamics Theory presented by Thannhauser (2009). The two core components, "grief" and "relationship dynamics," are linked together to demonstrate the connection and infinite cycle between one's inner experience and outside forces. Losses described in this theory include (a) major losses (physical health), and (b) secondary losses (e.g., consequences from the major loss, including identity shift, control, friends, hope for the future, normality, and assumptions about the world). The author also described manifestations of grief, or the emotional, social, cognitive, and behavioral response to loss reported by the adolescents with MS in her study. Denial was a distinctive reaction and possibly reflective of the developmental period. Acceptance followed grief and loss, and is described as a stage where the illness is integrated into identity development and coinsides with improvement in coping skills and mood. The second core component, 'relationship dynamics', is intertwined with grief, representing the force peer relationships have on change and well-being. Six subcategories were identified in Thannhauser's (2009) study that describe ways in which peer relationships connect to the grief experience: (a) medication-peer tug of war, (b) finding supportive relationships, (c) dealing with others' worry, (d) talking about MS, (e) acting normal, and (f) shifting friendships. Thannhauser's model of Grief-Peer Dynamics (2009) offers a

framework that can be applied to the experiences of emerging adults with JOMD. It captures the numerous losses driving a cycle, between grief and acceptance, that interferes with and complicates the developmental process. The subcategory of medication-peer tug-of-war does not apply to the JOMD population, as for them there are no injections or treatments that require extensive time and separation from peers. Emerging adults, not adolescents, were interviewed for this study, so romantic relationships and bearing and raising children were important relationships not addressed in Thannhauser's model, but likely to also contribute to the grief-peer dynamics.

Implications of Emerging Adults With JOMD for Counselor Education

Results of the present study have implications for counselors, counselor educators, and supervisors. The results represent a small sample of emerging adults living with JOMD, and as such, these recommendations serve as a starting point for further discussion of the mental health needs of this population. Implications for practice and education are further explored below.

Implications for Counseling Practice

As emerging adults with JOMD experience adjustment issues and difficulty coping, counseling may be warranted. It is likely helpful to use person-centered counseling services at various points as the illness progresses. The needs and goals of emerging adults with JOMD change over time and with progression of the disease. By and large, participants demonstrated ability to manage emotions independently. Traditional "therapy" for psychopathology, such as bipolar disorder or anorexia nervosa, is different from the counseling services being used to assist with illness related concerns.

This section provides recommendations for impactful and tailored individual counseling interventions for emerging adults with JOMD.

Individual counseling. Emerging adults with JOMD would benefit from individual counseling with encouraging, non-judgmental professionals are informed about JOMD. Typical stressors of the developmental period, such as change in school, job or housing, increase one's awareness of progression and cause more strain. Based on narratives of participants, this study concludes that emerging adults with JOMD would benefit from individual counseling services oriented towards assessing and attending to loss, facing fears, reorienting meaning, and nurturing identity development. Career counseling is another fundamental missing piece in comprehensive treatment that requires attention. Psychiatric consultation is yet another avenue that may assuage symptoms and increase one's sense of control. Finally, the researcher explores implications for counseling parents of children with JOMD.

Assess and attend to losses. Providing a safe space to delve into losses that have accompanied the diagnosis and progression of MD is a principal necessity. The MD impairments force young persons to transition from an able body to one with premature weakness. Counseling can help individuals process these untimely bodily changes, improve awareness of feelings, and coping with loss and protracted grief. Surprisingly, there is very little research on grief as it relates to progressive physical illness, and even less as it relates to the pediatric population. Nonetheless, it is a central source of emotional pain among this sample.

Individual counseling might begin with inquiry into the progression, stress and visibility of JOMD. For example, a counselor might ask, "How do you notice you live with MD? What kinds of things do you have trouble doing because of MD? What is that like for you?" Asking questions directly about loss must be done with an established rapport, focused presence, and gentility.

The graph utilized to depict Desmond's experience may also be useful while individually counseling clients. An adjusted version of the graph can be seen in Figure 6, JOMD Sample Progression Sequence. The graph on the right reflects the version to give to a person with JOMD to chart personal perception of stress and progression, seen in the figure on the left. The visibility barrier remains unchanged, but the progression and stress are unique to each person. The graph could be used as both a way to understand the distinctive experience of each person, track the perception of stress, and to normalize and talk about loss. Counselors might invite emerging adults with JOMD to chart their personal progression/losses and perceived stress. For example,

> JOMD begins as mostly invisible, and over time it becomes more noticeable, represented by the dotted line (on the graph). Please draw a (black) line that represents how JOMD has progressed over time, or gotten worse as you have grown up. Now, would you draw another (gray) line to depict how stressed you have felt, on average, over the years. Include times you have felt sad or anxious.

The results serve as a rich source of conversation, processing, and increase emotional awareness.

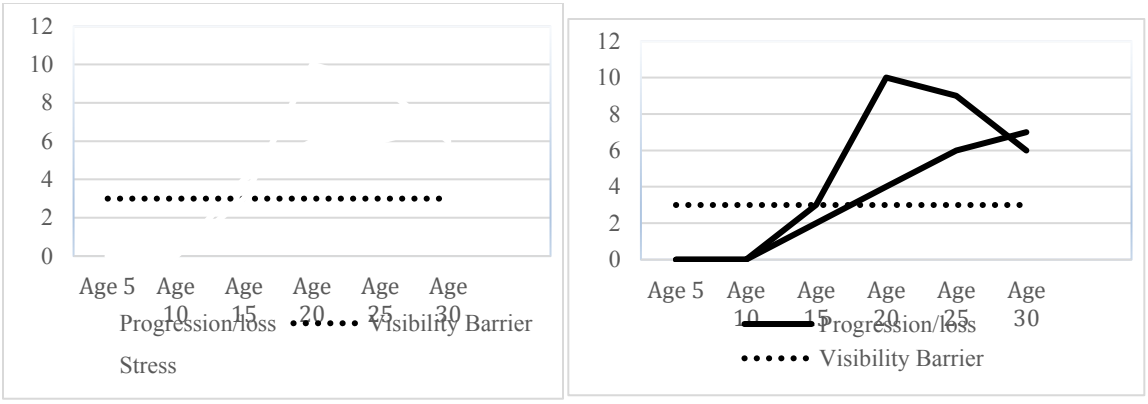

Figure 6. JOMD Sample Progression Sequence

The untimely nature of muscle loss, and/or cardiac and pulmonary health impairments, and subsequent forced transitions was, at times, severely distressing for participants. In terms of adolescent development, the losses those with JOMD experience come at a time when the brain regions for emotion regulation are still in the developmental process, and can be overwhelming for the patient to process (Feldman, 2011). The restoration orientation of the Dual Process Model (Stroebe & Schut, 1999) offers some insight into the denial and escapism behavior reported by participants in this study. In other words, the chronic grief that occurs because of living with JOMD requires times of moving away from the illness and negative emotions that it brings. The restoration orientation provides a period of protection from grief and feelings that are associated with it needs to be respected and honored by not pushing the patient to talk about JOMD too much, too fast.

Current theories on loss and grief have implications for counseling practice that apply to the JOMD population. Whether utilizing the Dual-Process Model or Meaning Reconstruction approach, both imply a non-linear path of processing grief, whereby

progress unfolds as the client vacillates between a loss and restoration orientation (Stroebe & Schut, 1999). Counselors can engage and facilitate growth for those with JOMD utilizing either model.

Results from this study suggest that it is normal for those growing up with JOMD to experience periods of intense grief. When the client engages in his or her losses, it is important to stay with these feelings and facilitate expression of the loss (assuming they are not self-harming) through verbal and nonverbal means. A counselor might ask a client, "What has been the hardest part of living with MD?" and await a story of loss. Be attuned to negative and positive narratives that may suggest healthy or unhealthy ways of thinking. Creative techniques, such as using a Styrofoam cup to represent a "healthy" body and then using a pencil to poke holes or otherwise mark the cup to symbolize losses because of JOMD, can bring life to emotions that need to be expressed. Drawing or sculpting feelings are other art therapy means of expression (Lister et al., 2008).

Emerging adults living with JOMD experience personal losses that propagate a restoration orientation. Counselors can help stimulate coping and problem solving, by role-playing how to tell someone about the condition in various possible scenarios, such as when help or accommodations are needed, or when informing a friend or possible romantic partner. Narrative therapy aims to separate the person from the problem, and that perspective is useful with this population. An intervention might include deconstructing the problem by talking about (or drawing/painting) parts of life that have become problematic because of JOMD (White, 2007).

Explore justification of JOMD and reconstruct meaning. With the loss that comes with JOMD, so does the opportunity for important learning experiences. It is through tragedy, death, and other traumas that people are forced to take on new perspectives in life that enhance well-being, empathy, and purpose (Middleton, 2016). As symptoms progress and patients become more conscious of living with JOMD, counselors have the potential to explore the justification process and facilitate 'meaning reconstruction' with their clients. Meaning reconstruction is a process whereby the traumatic circumstance is reinterpreted, or "made sense of," to establish a positive self-identity in the aftermath of a major personal loss (Neimeyer, 2000). An emerging adult with JOMD is faced with a collision between previously held goals and assumptions about life and specific stressful events, such as physical limitations requiring the use of an assistive device. One's personal experiences slowly diverge from that of a typical person in their 20s, a realization that deeply disrupts the life story. To grieve is to make sense of "why" this illness is a part of life, and reinterpret commonly held assumptions that disability is a tragedy. Finding benefits in the circumstance is part of reconstructing a new life story that incorporates the loss with positivity and resonance (Sapey, 2004).

Postmodern approaches are a natural fit for clients with JOMD. Logotherapy focuses specifically on the meaning one intends to fulfill in life despite tragic circumstances (Frankl, 1946; Zeligman et al., 2018). Narrative therapy also lends itself well to meaning reconstruction because it puts the client in an active position to reflect upon the past and intentionally create the future. In this study, participants reported

finding meaning by overcoming obstacles and fears, such as travelling to a new place or becoming a parent. Meaning was also derived from social interactions with friends and family, and a current, or hoped for, prospective career.

Reconstructing meaning is one way to personally develop after a life-altering event, promoting posttraumatic growth in individuals with chronic illness. Otherwise termed "existential growth" or "thriving," posttraumatic growth is a cognitive process that promotes growth through suffering (Middleton, 2016). Scholars have begun to examine posttraumatic growth for individuals living with various chronic illnesses, such as stomach cancer (Sim et al., 2015), chronic fatigue syndrome (Arroll & Howard, 2013) and HIV/AIDS (Amos, 2015), but none have examined this path for individuals living with neuromuscular disease. Standing in opposition to the traditional role of a sick person passively depending on healthcare professionals to manage chronic illness, those who have endured threats to psychological survival are willing and able to manage their own conditions as partners with professionals (Middleton, 2016). It is important for counselors to consider the meaning that can be made from a traumatic experience, as opposed to focusing mainly on the adverse impacts, especially for those with JOMD, because to be educated on the growth that can occur because of difficult circumstances might increase positive adjustment for the patient.

Face fears. To be an emerging adult with JOMD, is to be more aware of muscle deterioration since age of onset and, at times, it means fearfully contemplating how much worse the condition will get. In other words, what losses are yet to come? Counseling is an ideal place to explore anticipatory angst, especially as it relates to fears about future

progression and dependency. Inquiring specifically about fears because of living with JOMD, gives clients a chance to contemplate and express worries and consider what to do if/when those worries come to fruition. Upon building rapport, asking, "What fears do you have about living with JOMD?" provides an opportunity to further understand the client's perceptions of the illness and is consequences.

Results from this study and previous research (Aho et al., 2018) suggest that young persons with LGMD fear eventual wheelchair use. Rational emotive behavioral therapy might be useful in addressing this and other fears by identifying irrational beliefs that lead to distorted perceptions (Malkinson, 2010). Beliefs are irrational or dysfunctional if they are absolutistic evaluations that life events ought or must be different from the way they are, resulting in emotional distress (e.g., "I will have given up on myself if I start using a wheelchair;" "My life is worthless because I have JOMD;" Malkinson, 2010). On the other hand, rational or functional beliefs are realistic appraisals of adverse events based on preferences and acceptance (e.g., "There are aspects of being in a wheelchair that are difficult but it doesn't mean I am incapable;" "how unfortunate that I inherited this condition; my life may not be as I hoped it would, and it is sad and painful"). It can be difficult and anxiety provoking to consider a future with limited mobility, but taking the time to process fears can then empower a person to move forward and plan accordingly. Kwakkenbos et al. (2014) described interventions for targeting fears and uncertainty for an individual with progressive rheumatic disease. These interventions include exposure to thoughts about the future, education on functional versus dysfunctional fear, and restructuring strong cognitions such as how

much the illness impacts other parts of life. All those interventions, plus angst about possible dependency and rejection, would apply to emerging adults with JOMD.

Most emerging adults in the second decade of life consider a future with unlimited possibilities. How might their perspective on life change if they were faced with unavoidable muscle loss and disability? The notion of hopelessness is an important psychological symptom that may go undetected in the JOMD population, but has significant influence on health and well-being. Counselors must be sure to assess for hopelessness about the future when working with adolescents and emerging adults with JOMD.

Fears about being judged by others because of having JOMD, is another area for intervention. It is hypothesized that a main irrational belief dominating the narratives of participants was: "I am not as good as others, because I have JOMD," resulting in self-devaluation and emotional consequences of depressive mood and low self-esteem. In this instance, helping clients become less rigid and extreme through education about JOMD and its emotional consequences, in addition to developing more functional and creative ways to see oneself (e.g., "Many people eventually lose mobility in old age, maybe others will be more understanding than I thought") may be helpful. Teaching self-compassion and mindfulness may help those with JOMD cope with perceived shortcomings and develop strategies to comfort oneself in stressful moments and with feelings of failure (Neff, 2009). People who perceive themselves as worthwhile and happy feel more capable of change. Facing fears that come along with JOMD about

oneself, the world, and life itself is a challenge that emerging adults living with neuromuscular disease must deal with.

Nurture identity development. Living with JOMD imposes challenges to identity development. The degree of physical limitations in adolescence and young adulthood impacts one's ability to have new experiences, such as joining the basketball team or going on a mission trip. Lindsey articulated feeling "held back" in life, wishing she could participate in community sports and fundraising walks or runs. These restrictions are obstacles in exploring the self and discovering one's unique capabilities, delaying a fully formed identity. It is important, for improving quality of life, to recalibrate interests so that they are compatible with one's condition. Bishop's (2005; Bishop et al., 2007) Disability Centrality Model highlights the importance of adjusting the importance of the areas of life that are altered because of the chronic illness (Bishop, 2005). Counselors must be prepared to encourage an illness-identity that incorporates MD. Both narrative therapy and the social constructionist approach provide counselors with useful information and tools. It then becomes important to identify significant negative storylines stemming from distressing life events that limit and inhibit personal growth and to separate them from the individual, with the intention of "rewriting," to move from the known (problem story) to the unknown (White, 2007). Both the Dual Process Model and Meaning Reconstruction approach focus on the importance of reworking identity in grief work (Lister et al., 2008). While adolescence and emerging adulthood is a time of identity exploration, those living with JOMD require additional creativity and stress

tolerance to renegotiate personal, social, and professional status alongside progression of the disease.

Results of this study demonstrate that living with a progressive muscle disorder has psychological consequences, or common psychological states that accompany JOMD for emerging adults. If periods of suppression, anxiety, depression, and fatigue could be considered normal by-products of having this condition, it may reduce stigma and bring attention to treatable aspects of an otherwise untreatable illness. Those with JOMD were weary of burdening their support system with psychological problems, or judged themselves for being emotionally distressed. Integrating mental health—and possible psychiatric care or pharmaceutical assistance as part of the treatment for JOMD could potentially reduce distress and promote problem-solving behaviors.

Career counseling. Emerging adults living with JOMD need support from knowledgeable and sensitive professionals in providing career guidance. Career development is an important task in emerging adulthood, and serves as a buffer against MD symptoms that lower quality of life, such as social disengagement (Arnett, 2004; Pinquart, 2014). It is important to assist this population in finding a suitable occupation that reflects personal values, interests and considers current and pending physical limitations. Having an MD diagnosis restricts, to some degree, what vocational opportunities are available and adds to future uncertainty. Hoped-for options may not be possible with growing physical impairment, as was evident for several participants in this sample. For example, Sam was forced to drop out of automotive school because the physical demands were accelerating muscle breakdown. Desmond hoped to be a lab

technician, and now wonders if he would have the strength to reach and carry necessary equipment. These results are consistent with previous studies documenting the lower rates of young adults with chronic illness establishing employment (Pinquart, 2014). At a time when those with JOMD are experiencing more physical progression, career opportunities are also threatened. It is for this reason that career counseling would be beneficial in searching out other options.

Little literature exists exploring career counseling with young persons with juvenile onset chronic disease. Emerging adults with JOMD are ready and willing to work, but are concerned about aspects such as mandatory travel, inaccessibility of the workplace, and stigma from co-workers, all aspects to consider when utilizing career counseling with this population. It is for this reason the Work-life fit model (Moen, Kelly, & Huang, 2008) should be considered for career counseling those with JOMD, and possibly their—parent or spouse—caregivers (Hilbrecht et al., 2017). This model theorizes that the right fit, or the perception of having sufficient resources to function effectively both at work and at home, shifts over the lifespan as resources and needs evolve (Moen et al., 2008). One's career is a dynamic process that shifts with time and is largely dependent on perceived control of the following key resources: (a) Time for oneself, family, work and community; (b) sufficient income; (c) a work schedule that allows other responsibilities to be fulfilled; and (d) a feeling of job security (Hilbrecht et al., 2017). JOMD requires substantial reconfiguration of previously held assumptions about oneself and the future at various times throughout the lifespan, which aligns well with a model that clearly identifies change over the life course and poor health and

well-being of anyone, with or without disabilities, in a poor work-life fit. The benefits of periodic career counseling and needs assessments for emerging adults with neuromuscular disease is an investment in health and perceived well-being.

Psychiatric consultation. When working with this population, pharmacologic interventions should be considered in treating symptoms of depression, anxiety, and fatigue. First and foremost, a ruling out of another medical cause for symptoms, such as low vitamin D or thyroid dysregulation, is important in the medical treatment of any psychiatric disorder. Medical clearance provides opportunities for selective serotonin reuptake inhibitors (SSRI), selective norepinephrine reuptake inhibitors (SSNI) or stimulants (e.g., Ritalin) to improve symptoms of depression, anxiety, and lethargy. Combined with counseling services, medications are an integral component of evidenced based practice and recently documented in a guide to treatment of Duchenne MD (Birnkrant et al., 2018). There are, to date, no medications to help with disease progression, but secondary problems from the illness are treatable with medications that may offer some control in symptom management.

Counseling for Parents of Children With JOMD

How a parent responds to symptoms experienced by their child has a strong emotional imprint on participants, and in some ways, teaches the affected child how to react. For example, one participant reported feeling shame and doubt when she fell, partly because her mother would panic and shout when she was on the ground, reinforcing negative beliefs and increasing overall anxiety and depressive symptoms by her second decade. It is clear JOMD adds stress to the parent-child relationship, and

some families need education about the diagnosis, emotional support, and help in communicating.

Counselors could serve an important role in helping parents in the initial adjustment process by providing education to help them fully grasp the diagnosis and explore worries about future progression through individual or couples counseling (Aho et al., 2017). Parents and affected youth in this study were unprepared for initial physical changes. Instructing parents on what to expect in the first years of living with JOMD—including occasional falls, inability to run, difficulty climbing stairs—and what to do in those situations, may significantly reduce tension and improve the child/adolescent's ability to understand the disease. Counselors are skilled interviewers and could facilitate cathartic conversations among parents about their child's diagnosis, especially the hereditary aspect, which led some couples to blame each other. Parents mentally struggle when seeing their child physically struggle with the illness (Aho et al., 2017). Attending to parental distress begets opportunities to role-model empathic responding, that could carry over to the home environment. Ensuring mental health in parents sets the stage for cultivating a rewarding and validating home environment.

Previous research reveals that parents experience chronic grief in response to their child's condition, in addition to feeling unheard and under-valued by health care professionals (George et al., 2008). In addition to education on the illness and its effects, parents need a place to process the sadness and disappointment that comes along with having a child with JOMD and life turning out different from what was expected. Both individual or disease specific group therapy would be a useful adjunct to standard

treatment to get support and learn stress management. Several mothers in this study also served as caregivers for their young adult children, which magnifies stress in other areas of life, such as work and family responsibilities (Hilbrecht et al., 2017). Parents of young adults with LGMD2 reported a tendency to over-extend themselves, beyond their limits, in supporting and caring for their child (Aho et al., 2017). Interventions to help parents reconstruct meaning after the diagnosis and onset of the disease, through the sense-making, benefit finding, and identity change, could significantly accelerate awareness and growth (Gillies & Niemeyer, 2006).

In addition to education and attention to emotional distress, parents would benefit from learning how to talk about MD to the child who has it, siblings, medical professionals, and the community at large. We know from previous literature that parents are afraid to upset their child by bringing up the subject of JOMD (Aho et al., 2017), and the results of this study highlight that those who did not feel understood by mom or dad had a more difficult time adjusting and more expressions of anger. Several participants in the current study voiced their preference to protect their parents from worry about their health, and were withholding information about their physical and mental struggles. Others did not confide in their parents to avoid unwanted questioning. While no data was obtained directly from parents included in this study, it is evident that the parent-child relationship and the ability to communicate effectively among them, is a key influence in the lives of emerging adults with JOMD.

Family Systems Approach

Chronic childhood disease is not experienced in isolation. The economic, social, and psychological burden of pediatric illness impacts the entire family. Developmental difficulties and health problems extend beyond the identified patient to parents and siblings alike (Waldboth et al., 2016). Emerging adults with JOMD had ambivalent feelings towards their families. On the one hand, parents and siblings served as reliable caregivers, coaches, and allies supporting them in everyday life. But, on the other hand, tensions were reported between family members that caused frequent frustration and ongoing strained relationships. It is for these reason a family systems approach is suggested in the treatment of adjustment issues related to pediatric onset chronic illness (Waldboth et al., 2016) and JOMD, accordingly.

The family systems approach is a theory of human behavior whereby individuals are viewed as part of an emotional system; thus, problems are addressed with the context of the system, or family unit (Bowen, 1976). Members are systemically interdependent and linked in thoughts, feelings and behaviors, with problems being generated from issues with boundaries or identity (Bowen, 1976). Family members who have not yet had the chance to differentiate from the others are vulnerable to emotional distress (i.e., enmeshed boundary). JOMD impacts the entire family, thus it makes sense to consider this in best practices. The inevitable stress and anxiety that comes with this illness puts families at higher risk for dysfunction. The counselor's role is to help reveal the mechanics of dysfunctional family process by asking questions, providing education on

dysfunctional dynamics, promoting differentiation and assertive communication (Millington, 2012).

Counselors as part of an interdisciplinary treatment approach. Previous literature has identified the value of integrated treatment of chronic disease (Wu & Green, 2000). Yet, the counseling profession is undervalued and absent from interdisciplinary teams. Counselors can help identify the mental health needs and provide helpful, research based, interventions for individuals, families and couples. There is opportunity for medical professionals to enrich their knowledge and perspectives, by including professional counselors in their treatment of JOMD.

Counselors as advocates and informants for schools. School plays a critical role in social and cognitive development, especially during adolescence, and those with JOMD have special needs that may not be initially apparent to educators and staff. Counselors, either those employed by the school or professionals working in the community, are in an informed position to therapeutically ascertain delicate information about the physical needs and possible concerns for youth with MD, and discern how to best help the student minimize illness related stressors during the school day. Since the condition is degenerative, it would be ideal for school counselors and teachers to be updated at least once a year and to expect growing needs. Simple accommodations, such as a heavy table and chair instead of an individual desk, or teachers hand-delivering homework/tests instead of calling the student to stand and retrieve it, may significantly reduce distractions and unnecessary worry. Results from this study suggest that general education about MD is needed among teachers and, possibly, peers, thereby reducing the

pressure on the student to explain and justify symptoms or accommodations. Licensed professional counselors can inform teachers and collaborate with school staff about the physical and emotional consequences of JOMD, especially regarding physical activities, like going outside or sitting on the ground where it might be difficult for the student to get back up. Advocating for the student with JOMD must be balanced with respect for privacy and need for self-sufficiency.

Implications for Counselor Education and Supervision

Results of this study have implications for those engaged in the pedagogy and supervision of counselors, both before and after graduation.

The mental health field may be appealing for those living with JOMD, as it does not have physical requirements and provides the opportunity to help others. Students with JOMD offer a unique perspective on loss and suffering. Nonetheless, understanding the struggles these students face would be useful in the educational context. Asking for accommodations in education is difficult for this population, but may be necessary in retention and graduation. For example, one participant dropped out of college because she could no longer get out of desks and chairs without assistance. Faculty addressing these worries with the student with JOMD, would provide a positive learning environment and decrease fears that the disability will limit success. It would also be important to consider the student's physical limitations in practicum and internship placement, ensuring accessibility and communicating possible needs to the site supervisor.

In terms of education and supervision, the results of this study have implications for the training of mental health-, rehabilitation-, and school-counselors. While educators cannot possibly keep track of the unique needs of each medical diagnosis, they can advocate and encourage students to turn to the literature when they have a client with a chronic illness. Furthermore, counselor educators can address the gap in treatment for those with disabilities and ensure graduates are sensitive to and informed about stigma and incorrect stereotypes. Results of this study could be helpful to counselor educators supervising a student who is counseling a person with JOMD. These cases are opportunities for the student to grow in learning about the psychological impact of MD. Finally, counselor educators might consider including the concept of posttraumatic growth when teaching about trauma, chronic illness and disability.

Workshop for Post-Graduate Training

Disseminating the information gathered from research may be best suited to a workshop or conference presentation. Those in the field of mental health and medicine may be interested in learning more about the experiences of young persons with slowly progressive MD. Below is a brief description of one possible seminar.

Workshop Proposal Title: Counseling adolescents and emerging adults with juvenile onset muscular dystrophy (MD)

Abstract: With 21st century advancements in medicine and technology, diagnoses of children with MD are occurring at a faster rate than ever before in history. Genetic developments have drastically improved the identification of various types of MD diagnosed in childhood, such as LGMD 2a, that do not alter life expectancy, but cause

significant physical disability over the lifespan. While there are currently no treatment options to slow or stop skeletal muscle, cardiac, and/or lung function deterioration, secondary symptoms such as depression, anxiety, low self-esteem, fatigue, and family stress are treatable. Counseling is an underutilized service in the treatment of chronic disease, but licensed professionals are valuable in addressing adjustment problems and coping issues. This workshop will inform counselors about MD, including the experiences of emerging adults who live with it, and educate attendees on professional implications. Young adults with JOMD and their families are greatly in need of counseling services to grieve losses, express fears, reconstruct meaning, and enhance career development.

Who should attend this workshop? Understanding the experiences of adolescents and emerging adults living with MD since childhood adds to the toolkits of those in the field of mental health and medicine. Licensed professional counselors, social workers, nurses, psychologists, psychiatrists, and medical doctors−especially those in family medicine, neurology, and palliative care−may benefit from this workshop.

What participants will learn during the workshop: Participants will learn more about types of MD diagnosed during childhood, specifically LGMD and EDMD. Common problems among adolescents and emerging adults with chronic illness will be explored. Developmental considerations of living with a progressive and degenerative condition that causes untimely losses will also be provided, in addition to specific counseling interventions that may be warranted during times of adjustment. Support from family, school, and pharmacology is discussed.

Transferability and Trustworthiness of the Data

In qualitative research, trustworthiness of data is established through a rigorous framework to accurately portray a true, or credible picture of the phenomenon being studied (Shenton, 2004). It is upon this foundation that transferability is considered. Specific contextual information, such as the number of participants, data collection methods, and length of interviews (see Chapter 2) provide the means for the reader to ultimately determine how much and in what situations the results and conclusions of this study are transferable (Lincoln & Guba, 1985). While on the one hand the findings are specific to this sample of emerging adults with JOMD, it adds to the accumulation of findings from studies staged in different setting (e.g., hospitals) to provide a comprehensive understanding of living with MD (Shenton, 2004). The procedures employed to increase trustworthiness and subsequent transferability for the results in this study include: (a) bracketing; (b) triangulation, or interviewing multiple emerging adults with JOMD; (c) strategies to promote honesty in participants for accurate and nuanced descriptions; and (d) member checks.

Bracketing was completed prior to data collection to capture and quarantine the researcher's experiences and inherent biases about living with JOMD. Bracketing and quarantine were utilized again during data analysis to sideline any personal intrusions or false judgments influencing the findings. The transcripts and transformations were returned to again and again and submitted to peer review to build the five themes developed; while the results do share similarities with the researcher's experience, the findings are constructed from the participants' experiences.

Interviewing eight participants on two separate occasions demonstrates triangulation, or the representation of multiple, diverse viewpoints regarding the same experience (Shenton, 2004). Individuals from the United States lived in Ohio (3), Florida (1), New York (1), Pennsylvania (1) and Massachusetts (1). One participant resided in Germany. The intentional, purposeful, sampling of emerging adults living with specific types of MD that are slowly progressive, increases trustworthiness of the findings for the phenomenon under investigation; and within the sample there is variation among the types of MD, gender, and age.

Thick, rich descriptions help to convey the experience and provide readers the opportunity to relate to the findings, hence increasing transferability. To promote honesty, the researcher provided opportunities for participants to refuse answering questions, or stop the interview at any point. The researcher aimed to establish rapport at the onset of each interview and emphasized that there were no right or wrong answers. Participants were encouraged to be direct and honest, and only take part in the research if they were genuinely willing and prepared to share their experiences.

An important element to establishing trustworthiness of the findings is to confirm that the raw data is, indeed, what was reported during the interview and that the themes "ring true" for each participant (Merriam, 2002). Participant checking in this study occurred on three separate occasions: (a) participants read the first interview transcript and verified that it reflected what they intended to communicate about the experience; (b) during the second interview, when key points from the first dialogue were summarized by the researcher and confirmed by the participant; and (c) emailing the themes to each

person in the sample with the request to confirm the findings. Ensuring accurate data from informants is considered one of the most important methods for establishing trustworthiness (Lincoln & Guba, 1985; Shenton, 2004).

Phenomenologists seek to determine what an experience means for the persons who have had the experience, and from those individual descriptions develop an integrated synthesis that portrays the essence of it (Moustakas, 1994). The researcher hopes that the results of this study will resonate with and be of value to others living with JOMD. The descriptions are also intended to educate people with little or no knowledge of the phenomenon what it is like to live with a slowly progressive muscle disease in emerging adulthood. At any time, these people may get confronted, professionally, personally, or socially with emerging adults with JOMD.

Limitations

This phenomenological inquiry had several limitations. Methodological issues, such as data sources, unequal representation of JOMD types, demographics of participants, and researcher self-disclosure are specific limitations that relate to the transferability of the research.

Difficult decisions about inclusion criteria and recruitment techniques were made at the onset of the study, and were adjusted to accommodate the unexpected difficulty obtaining participants. To maximize access to data sources, multiple sources of recruitment were utilized, including two key informants (one in Ohio and another in Arizona) who personally contacted over a dozen potential participants. The other access portal included five MD specific Facebook pages. The researcher also contacted the

MDA national headquarters for assistance, but the organization was unresponsive. As such, only those who had a connection to the key informants or were members of the Facebook groups where the study advertisement was posted had access to the research. Despite multiple posts, no person with BMD or FSHMD volunteered to be interviewed, leaving only two types of MD represented in this sample, unequally at that, as seven of the eight participants had LGMD. Thus, findings are mostly a reflection of the experiences of emerging adults with LGMD. Finding participants who met the study criteria and were willing to be interviewed proved to be a difficult task and reflects a limitation.

Sixteen interviews took place from August 2016 through September 2017 to gather extensive descriptions of the experience of living with JOMD from eight participants. Missing data, including a second interview transcript and missing nonverbal cues that cannot be captured via telephone, are further limitations of the results. A recording failure led to the loss of data and subsequent transcript with Sarah. While the researcher recorded a detailed review of the interview, missing verbatim data is a deficit in the results. Important information about her individual experience and its impact on the collective narrative is missing. Furthermore, six of the eight interviews occurred over the telephone. Had the researcher had the opportunity to gather data in person, nonverbal communication—such as facial expressions, silent tears—could have added to the results.

Personal experience with the topic under study may have influenced the data collection and analysis process. The researcher self-disclosing her and her brother's diagnosis of LGMD2a and minimal private details (e.g., age diagnosed, current physical

mobility) was intentional and served as time invested in establishing rapport at the beginning of the interview. If a phenomenological researcher wants a deeply revealing experience to be articulated, a relationship greater than that of a passing acquaintanceship is required (Giorgi, 2009). This approach was useful and facilitated disclosure by participants. In fact, half of participants were delighted because the researcher was the first and only person, they had ever spoken with, who also lived with JOMD. At the end of his second interview, Sam described his feeling of inspiration through telling his story and learning of the researcher's efforts and interest in JOMD.

> I'm really happy that there is other people out there that are pushing just as hard, and realize that it's (EDMD) not just a normal condition, even though you get to live to be 80 or 90 or whatever; it's not just a thing you can brush under the rug. I hope everyone else you got to speak with . . . got the same aide out of it from finally talking about it.

By sharing brief details of personal history, the researcher established a field of intimacy and psychological sensitivity to the topic.

Personal history and experience of the researcher may also have influenced the data analysis process. The researcher living with JOMD herself, may have sensitized her to certain topics. Attending to some aspects of the data, while dismissing others, are necessary decisions to filter down data to purposeful statements in qualitative research (Schram & Schram, 2006). This was done through several steps. First, only the individual meaning statements related to the experience of living with JOMD were included. Next, statements that were similar in nature were grouped together under an

initial theme name, such as "Emotional Impact" and "Progression and Accepting Help," and organized chronologically in the transformation chart. Third, the transformation charts were utilized to create the integrated narrative, which summarized the narrative of each participant. Finally, transformation charts and integrated narratives of all the participants were compared, grouping similar data across participants together, which, eventually, comprised five themes representing the essence of the experience. To minimize potential bias, the interview transcripts, transformation charts, and final themes were submitted to a peer auditor to confirm neutrality of the finding. During the analysis transformation and analysis process, the researcher returned to the data time and again to ensure the results were an accurate reflection of the interview data itself (Giorgi, 2009).

Recommendations for Future Research

The results of this study offer opportunities for future research, several of which are generated from the limitations. First, a larger scale study assessing the experiences of emerging adults living with various forms of JOMD is necessary, especially in the United States, as most the psychosocial literature has taken place abroad. Broadening the inclusion criteria to other forms of neuromuscular disease diagnosed in childhood, such as Fredrick's ataxia or spinal MD, would offer deeper understanding. Collecting data in person or in focus groups next time would also enrich the data and test the findings of this study. Separating sub-types and researching one type of JOMD at a time—such as EDMD or Becker MD—would clarify similarities and differences within the MD community.

As results here suggest, living with JOMD is emotionally taxing and frightening. At present, we have no data on what the rate of suicide ideation and completion is among the MD population. Completing an emotional impact study, including gathering information about suicidality, depression, anxiety, and fatigue would further assist in identification of needs and helpful interventions. Another idea is to separate the sub-themes into individual studies (JOMD related depression, anxiety, etc.) to clarify the dimensions and variations of the concepts articulated here. More information, on a larger scale, is needed about the emotional consequences of living with JOMD.

Research opportunities also exist within the implications and recommendations for counseling. Research, specifically investigating the counseling experiences of those with MD, would provide first-hand accounts of what initiated the use of counseling services and what was most beneficial in treatment. Such a study could also inform mental health providers of problems that lead to early disengagement in mental health treatment. By studying the counseling experiences of those with JOMD, helpful information about the most stressful times and aspects of effective therapeutic interventions, as well as career counseling needs, could be identified.

Interviewing adolescents who live with JOMD would provide further information about school and home obstacles. Utilizing the Peer-Grief Dynamic model as a lens, future research could identify first and second-degree losses of JOMD, and turning points of adjustment (Thannhauser, 2014).

Finally, meaningfulness is emerging as a key concept in transcendence—or positive adjustment to disability. With a body that continually changes, so does the

ability to participate in certain meaningful activities. Long-term research that follows individuals with MD throughout the lifespan and compiles data on meaningful activities would be helpful.

Summary of Chapter IV

Results from this qualitative inquiry add to the much-needed psychosocial literature on JOMD. Several significant findings were discussed. The losses that emerging adults with JOMD experience lead to changes in consciousness as one navigates developmental milestones while physicality is continually worsening. This experience aligns with theories on grief, a concept missing from the MD literature. The cognitive efforts required to process the losses and make adaptions to decreasing physical strength over time increase stress and therefore, susceptibility to high stress and mental or emotional disorders. By normalizing grief as part of the JOMD experience, individuals and families can have a better understanding of what to expect, and learn to create new meaning and appreciation in life. Another contribution was the perceived stigma during the transition from invisible to visible symptoms, and how this proved to be an obstacle in accomplishing developmental milestones. The way in which emerging adults "talk to themselves" about the illness also plays an important role in adjustment. Finally, while not a new contribution to the literature, this study emphasizes the negative emotions and fear of the future that occur with JOMD. Existential questioning and hopelessness elicited suicidal ideations among some participants.

The implications for counseling practice and education were discussed. Interventions for emerging adults with JOMD, such as assessing and attending to losses,

exploring how one is justifying JOMD, reconstructing meaning, facing fears, nurturing identity development and career counseling offer a first attempt to provide more comprehensive and helpful treatment. Interdisciplinary care, such as working with psychiatrists and schools, may also improve quality of life. Finally, it is important to not only treat the patient with JOMD, but also the family (especially parents) who also play an important role in disease management. Counselor educators and supervisors would benefit from understanding the experience of JOMD with affected students, supervisee's or clients. Limitations of this study, including the researcher herself having JOMD and the uneven sample composed mostly of the LGMD type were discussed. Finally, recommendations for future research opportunities, such as assessing suicidality, were explored.

Conclusion

The purpose of this study was to understand how emerging adults perceive and describe their experiences of living with JOMD. Most of the extant literature on MD has focused on physical symptoms. Consequently, there are gaps in the knowledge relating to emerging adults' experiences of living with JOMD. Research is needed to enhance our understanding of youth living with slowly progressive neuromuscular disease. The findings of this study suggest that emerging adults living with a juvenile onset form of MD face unique challenges that interrupt psychosocial development. Living with JOMD is a complex experience that requires a multidisciplinary approach, including counseling, to offset the challenges. The results of this study lead us to suggest that in managing JOMD, the assessment and detection of the presence and severity of associated

psychological distress may provide dramatic improvement in quality of life and adjustment in emerging adulthood. Counseling may make the difference between adjustment and serious psychological problems that delay development.

APPENDICES

APPENDIX A

KENT STATE UNIVERSITY INSTITUTIONAL REVIEW BOARD APPROVAL

Appendix A

Kent State University Institutional Review Board Approval

Hello,
I am pleased to inform you that the Kent State University Institutional Review Board reviewed and approved your Application for Approval to Use Human Research Participants as a Level II/Expedited, category X project. **Approval is effective for a twelve-month period:**

June 17th, 2016 through June 16th, 2017

**If applicable, a copy of the IRB approved consent form is attached to this email. This "stamped" copy is the consent form that you must use for your research participants. It is important for you to also keep an unstamped text copy (i.e., Microsoft Word version) of your consent form for subsequent submissions.*

Federal regulations and Kent State University IRB policy require that research be reviewed at intervals appropriate to the degree of risk, but not less than once per year. The IRB has determined that this protocol requires an annual review and progress report. The IRB tries to send you annual review reminder notice by email as a courtesy. **However, please note that it is the responsibility of the principal investigator to be aware of the study expiration date and submit the required materials.** Please submit review materials (annual review form and copy of current consent form) one month prior to the expiration date. Visit our website for forms.

HHS regulations and Kent State University Institutional Review Board guidelines require that any changes in research methodology, protocol design, or principal investigator have the prior approval of the IRB before implementation and continuation of the protocol. The IRB must also be informed of any adverse events associated with the study. The IRB further requests a final report at the conclusion of the study.

Kent State University has a Federal Wide Assurance on file with the Office for Human Research Protections (OHRP); FWA Number 00001853.

If you have any questions or concerns, please contact the Office of Research Compliance atResearchcompliance@kent.edu or 330-672-2704 or 330-672-8058.

APPENDIX B

SECOND IRB APPROVAL

Appendix B

Second IRB Approval

Hello,
The Kent State University Institutional Review Board (IRB) has reviewed and approved your Annual Review and Progress Report for continuing review purposes. The protocol approval has been extended and is effective**:**

May 1, 2017 through April 30, 2018

For compliance with:

- DHHS regulations for the protection of human subjects (Title 45 part 46), subparts A, B, C, D & E
-

**A copy of the IRB approved consent form may be attached to this email if the study is still recruiting in person. This "stamped" copy is the consent form that you must use for your research participants. It is important for you to also keep an unstamped text copy (i.e., Microsoft Word version) of your consent form for subsequent submissions.*

Federal regulations and Kent State University IRB policy requires that research be reviewed at intervals appropriate to the degree of risk, but not less than once per year. The IRB has determined that this protocol requires an annual review and progress report. The IRB will try to send you an annual review reminder notice by email as a courtesy. **However, please note that it is the responsibility of the principal investigator to be aware of the study expiration date and submit the required materials.** Please submit review materials (annual review form and copy of current consent form) one month prior to the expiration date.

HHS regulations and Kent State University Institutional Review Board guidelines require that any changes in research methodology, protocol design, or principal investigator have the prior approval of the IRB before implementation and continuation of the protocol. The IRB must also be informed of any adverse events associated with the study. The IRB further requests a final report at the conclusion of the study.

Kent State University has a Federal Wide Assurance on file with the Office for Human Research Protections (OHRP); FWA Number 00001853.

APPENDIX C

RECRUITMENT EMAIL (INITIAL)

Appendix C

Recruitment Email (Initial)

My name is Kelsey A. Jager, and I am a Ph.D. candidate in Counseling Education and Supervision at Kent State University. I am conducting a phenomenological study examining the experiences of emerging adults (aged 18 to 29) living with a slowly progressive form of Proximal Muscular Dystrophy (termed Juvenile Onset). I'd like to interview individuals who have been diagnosed in childhood with forms of the disease such as Limb Girdle, Distal, Beckers, Facioscapulohumeral and Emery-Dreifuss (not the Duchenne or Congenital forms). Jason McGlothlin, Ph.D. and Marty Jencius, Ph. D are my dissertation co-chairs, and this study has been approved by the Kent State University Institutional Review Board.

I am writing to ask for assistance in recruiting individuals willing to participate in my dissertation study investigating the experiences of emerging adults (aged 18-29) living with juvenile onset muscular dystrophy. I hope to better understand how emerging adults perceive and describe their experiences of living with a form of muscular dystrophy diagnosed in childhood, and how it affects life during emerging adulthood.

Participants in the study must be between the ages of 18 and 29 and have a documented diagnosis of muscular dystrophy that is not the Duchenne type. The diagnosis of Muscular Dystrophy must be received at least one year prior to participation in this study. Participants need to be able to reflect on and comprehensively describe their experiences of living with the disease. Thus, they must have no known cognitive impairments or problems understanding English that would impair his or her ability to participate in the interview. Finally, participants must be willing to participate with two 60-90 minute interviews.

I would sincerely appreciate any help you might be able to provide in the form of recommending individuals you know

If you are able to help, please forward this email to potential candidates, or provide me with names and contact information of individuals who might be willing to participate. Please contact me at kgeorge1@kent.edu or (440) 554-4668 to let me know if you would be willing to assist me, or for more information.

Thank you for your time, and I look forward to hearing from you.

Best Regards,
Kelsey A. Jager
Doctoral Candidate
Kent State University

APPENDIX D

RECRUITMENT EMAIL REVISED

Appendix D

Recruitment Email Revised

To Whom It May Concern:

Thank you for assisting me in my study, titled *The Experiences of Emerging Adults with Juvenile Onset Muscular Dystrophy.* Please forward this email, or copy and paste the entire message below to your listserv so that I might recruit young adults living with muscular dystrophy. Please do not alter the description below.

You are invited to participate in a research study examining the social and emotional aspects of living with Muscular Dystrophy as a young adult. This is a qualitative investigation that will require between 2 to 5 hours of your time.

The researcher is recruiting individuals with Muscular Dystrophy who meet the following criteria:

- are between the ages of 18 and 29
- physical symptoms become noticeable after age 9 (termed Juvenile Onset)
- living with a slowly progressive form of the disease such as Limb Girdle, Distal, Becker's, Facioscapulohumeral and Emery-Dreifuss (not the Duchenne or Congenital forms)
- have been diagnosed with Muscular Dystrophy for at least one year prior to participation

The study commitment:

- Two 60-90 minute interviews within 2 weeks
- Interviews will be audio recorded and take place over the phone, in person or via Skype
- All information will be kept confidential and identifying information (e.g., name, where you live) will be changed in the final publication

You will be asked questions about being diagnosed with muscular dystrophy, and how it has affected your life socially and emotionally, how you cope, and what services might be helpful to you

You will be asked to review the first interview transcript to ensure its accuracy

Your participation in this study is completely voluntary, and you can withdraw at any time without penalty or loss of benefits to which you are otherwise entitled. Your participation (or lack thereof) will not influence services provided through the Muscular Dystrophy Association.

This research is being conducted to fulfill requirements for the degree of PhD in Counselor Education and Supervision at Kent State University Graduate School of Education, Health and Human Services in Kent, Ohio.

If you have questions or are interested in participating, please contact Kelsey Jager at kgeorge1@kent.edu or text/call (440) 554-4668.

This project has been approved by the Kent State University Institutional Review Board (approval number 16-297). If you have any questions about your rights as a research participant or complaints about the research, you may call the IRB at (330) 672-2704.
Thank you for your time, and I look forward to hearing from you!

Best Regards,
Kelsey A. Jager
Doctoral Candidate
Kent State University

APPENDIX E

FACEBOOK RECRUITMENT ADVERTISEMENT

Appendix E

Facebook Recruitment Advertisement

You are invited to participate in a research study examining the social and emotional aspects of living with Muscular Dystrophy as a young adult. This is a qualitative investigation that will require between 2 to 5 hours of your time.

The researcher is recruiting individuals with Muscular Dystrophy who meet the following criteria:
are between the ages of 18 and 29
physical symptoms become noticeable after age 9 (termed Juvenile Onset)
living with a slowly progressive form of the disease such as Limb Girdle, Distal, Becker's, Facioscapulohumeral and Emery-Dreifuss (not the Duchenne or Congenital forms)
have been diagnosed with Muscular Dystrophy for at least one year prior to participation

The study commitment:
Two 60-90 minute interviews within 2 weeks
Interviews will be audio recorded and take place over the phone or in person

All information will be kept confidential and identifying information (e.g., name, where you live) will be changed in the final publication

You will be asked questions about being diagnosed with muscular dystrophy, and how it has affected your life socially and emotionally, how you cope, and what services might be helpful to you

You will be asked to review the first interview transcript to ensure its accuracy

Your participation in this study is completely voluntary, and you can withdraw at any time without penalty or loss of benefits to which you are otherwise entitled. Your participation (or lack thereof) will not influence services provided through the Muscular Dystrophy Association.

This research is being conducted to fulfill requirements for the degree of PhD in Counselor Education and Supervision at Kent State University Graduate School of Education, Health and Human Services in Kent, Ohio.

If you have questions or are interested in participating, please contact Kelsey Jager at kgeorge1@kent.edu or text/call (440) 554-4668.

This project has been approved by the Kent State University Institutional Review Board (approval number 16-297). If you have any questions about your rights as a research participant or complaints about the research, you may call the IRB at (330) 672-2704.
Thank you for your time, and I look forward to hearing from you!

APPENDIX F

SCREENING AND DEMOGRAPHIC FORM

Appendix F

Screening and Demographic Form

The Experiences of Emerging Adults Living with JOMD

Name: _____

Address:
_____
_____
_____

Phone Number: _____

E-mail: _____

Date of Birth: _____
When were you diagnosed with Muscular Dystrophy?
_____

What kind of Muscular Dystrophy do you have? _____
Subtype? _____
Who diagnosed you? _____
Are you willing to participate in two interviews to describe living with muscular dystrophy, each about 60 to 90 minutes each? Yes_____ No _____

Date/time/location of first interview:
_____
_____
_____

APPENDIX G

PARTICIPANT CONSENT FORM

Appendix G

Participant Consent Form

Informed Consent to Participate in a Research Study

Study Title: *The Experiences of Emerging Adults with Juvenile Onset Muscular Dystrophy*

Principal Investigator: *Jason McGlothlin* **Co-Investigators:** *Martin Jencius, Ph.D.*
Kelsey A. Jager, M.Ed. PCC

You are being invited to participate in a research study. This consent form will provide you with information on the research project, what you will need to do, and the associated risks and benefits of the research. Your participation is voluntary. Please read this form carefully. It is important that you ask questions and fully understand the research in order to make an informed decision. You will receive a copy of this document to take with you.

Purpose:
The purpose of this phenomenological study was to understand the lived experiences of emerging adults (aged 18-29 years old) living with juvenile onset muscular dystrophy.

Procedures
Your participation will include meeting for two semi-structured interviews lasting approximately 60 to 90 minutes each via face-to-face, Skype, or telephone. The interviews will focus on your experiences of living with muscular dystrophy during emerging adulthood. You will be emailed or postal mailed a copy of the interview transcript and themes generated for member checking.

Audio Recording
Each interview will be audio recorded with a digital, portable device and transcribed for data collection and analysis. The recordings will be used for research purposes only, and deleted at the conclusion of the study.

Benefits
Although this research will not benefit you directly, your participation in this study will help us to better understand the experiences of emerging adults living with Juvenile Onset Muscular Dystrophy. Your participation offers the opportunity to reflect on and discuss your experiences of living with a slowly progressive form of Muscular Dystrophy with a mental health professional familiar with the disease.

Risks and Discomforts

You will be asked questions that require you to reflect on and discuss living with Juvenile Onset Muscular Dystrophy. The personal nature of the topic may cause you emotional distress. You may ask to see the questions before deciding whether or not to participate in the study.

Privacy and Confidentiality

Your study related information will be kept confidential within the limits of the law. Any identifying information will be kept in a secure location and only the researchers will have access to the data. Research participants will not be identified in any publication or presentation of research results. The consent forms will kept in a locked location in 310 White Hall, Kent State University.

Your research information may, in certain circumstances, be disclosed to the Institutional Review Board (IRB), which oversees research at Kent State University, or to certain federal agencies. Confidentiality may not be maintained if you indicate that you may do harm to yourself or others.

Voluntary Participation

Taking part in this research study is entirely up to you. You may choose not to participate or you may discontinue your participation at any time without penalty or loss of benefits to which you are otherwise entitled. You will be informed of any new, relevant information that may affect your health, welfare, or willingness to continue your study participation.

Contact Information

If you have any questions or concerns about this research, you may contact Kelsey Jager at (440) 554-4668, Jason McGlothlin at (330) 672-0716 or Marty Jencius at (330) 672-0699. This project has been approved by the Kent State University Institutional Review Board (insert approval number here). If you have any questions about your rights as a research participant or complaints about the research, you may call the IRB at 330.672.2704.

Consent Statement and Signature

I have read this consent form and have had the opportunity to have my questions answered to my satisfaction. I voluntarily agree to participate in this study. I understand that a copy of this consent will be provided to me for future reference.

_____ _____

Participant Signature **Date**

APPENDIX H

AUDIOTAPE CONSENT FORM

Appendix H

Audio Tape Consent Form

The Experiences of Emerging Adults Living with JOMD
Principle Investigator: Jason McGlothlin, Ph.D.

Co-investigators: Martin Jencius, Ph.D., & Kelsey A. Jager, M.Ed.

I agree to participate in an audio-taped/video taped interview about the experiences of emerging adults living with a form of juvenile onset muscular dystrophy as part of this project and for the purposes of data analysis. I agree that Kelsey A. Jager may audio-tape/video tape this interview. The date, time and place of the interview will be mutually agreed upon.

_____ _____

Signature Date

I have been told that I have the right to listen to the recording of the interview before it is used. I have decided that I:
____want to listen to the recording ____do not want to listen to the recording
Sign now below if you do not want to listen to the recording. If you want to listen to the recording, you will be asked to sign after listening to them.

Kelsey A. Jager may / may not (circle one) use the audio-tapes/video tapes made of me. The original tapes or copies may be used for:

____this research project _____publication _____presentation at professional meeting

_____ _____

Signature Date

APPENDIX I

PARTICIPANT DATA SHEET

Appendix I

Participant Data Sheet

How do you describe your nationality, race, and/or ethnicity?
_____

What is your gender? _____ Male _____ Female _____ Transgender _____ Other

What is your employment status? _____
If employed, what is your current occupation?
_____

What is your highest level of education? _____
If currently attending college, what is your major?
_____

What is your relationship status?
_____ Single
_____ Married _____ Month _____ Day _____ Year
_____ In a relationship _____ How Long?
_____ Divorced _____ Year of Divorce

Who lived in your house in your house at the time of diagnosis?
_____

Who lives with you now? _____
Pets? _____

APPENDIX J

INITIAL INTERVIEW QUESTIONS

Appendix J

Initial Interview Questions

The Experiences of Emerging Adults Living with JOMD

Qualitative Semi-Structured Questionnaire

1. How would you describe your experience of living with your condition, beginning with your first memories of the disease up until now?
    a. What aspects (being diagnosed, loss of ability, etc.), occurrences, or people connected to the experience stand out to you?
    b. How does having muscular dystrophy affect you and your life? How has it changed you?
    c. How do you believe having your condition has affected significant others in your life?
    d. Who, if anyone, do you lean on for support in coping with your condition?

2. What is the most difficult aspect of having this diagnosis as an emerging adult?

3. What, if anything, do you believe could be helpful to you to better cope with your condition?

4. How do expect your condition will impact your future?

5. How did you get more information about your diagnosis (from a parent, doctor, self)? When did you learn it? What was it like?

6. Have you shared all that is significant about your experience of living with this diagnosis?

APPENDIX K

SECOND INTERVIEW QUESTIONS

Appendix K

Second Interview Questions

The Experiences of Emerging Adults Living with JOMD
Qualitative Semi-Structured Questionnaire

1. What thoughts/feelings emerged for you as you reflected on our first interview?
2. What stood out for you the most from our first interview?
3. Has anything been excluded about your experiences of living with this diagnosis?
4. Have you shared all that is significant about your experiences of living with your condition?

APPENDIX L

MENTAL HEALTH REFERRALS

Appendix H

Mental Health Referrals

Cuyahoga County		
	Recovery Resources 14805 Detroit Ave, Suite 200 Lakewood, Ohio 44107 (216) 431-4131	Catholic Charities of Cuyahoga County 7800 Detroit Road Cleveland, Ohio 44102 (216) 631-3499
	Cleveland Clinic Foundation Department of Psychiatry and Psychology Desk P57 9500 Euclid Ave. (216) 636-5860	Community Behavioral Health 3355 Richmond Rd. Beachwood, Ohio 44122 (216) 831-1494
	Far West Center 29133 Health Campus Drive Westlake, Ohio 44145	Murtis Taylor Human Services Multiple locations in Cleveland, Ohio (216) 283-4400
Summit County		
	Coleman Professional Services 444 N. Main Street Akron, Ohio 44310 (330) 379-0667	Community Support Services 150 Cross Street Akron, Ohio 44310 (330) 253-9388
	Portage Path Behavioral Health 340 S. Broadway Street Akron, Ohio 44308 (330) 253-3100	Clinic for Individual and Family Counseling The University of Akron 27 South Forge Street Akron, Oh 44325 (330) 972-6822
Lorain County		
	Kenneth A. DeLuca, PhD & Associates, Inc. 35888 Center Ridge Rd., #5North Ridgeville, Ohio 44039 (440) 327-1800	Pathways Counseling & Growth Center 312 Third Street Elyria, Ohio 44035 (440) 323-5707

APPENDIX M

FOLLOW UP EMAIL

Appendix M

Follow Up Email

Dear Participant,
Thank you again for your participation in the study titled "*The Experiences of Emerging Adults with Juvenile Onset Muscular Dystrophy: Implications for Counselors."* The following are themes that emerged from my data analysis.
Theme 1: JOMD expands into consciousness with continued loss
Theme 2: Stress of experiencing increased visibility
Theme 3: Self-other justification of JOMD
Theme 4: Evolution of disease, evolution of negative emotion

Sub-themes included JOMD related suppression, anxiety, depression and fatigue
Theme 5: Fear of the future
Please take the opportunity to review the themes and provide any feedback. You may contact me via phone (440-554-4668) or email (kgeorge1@kent.edu). Thank you again for your time and consideration.

Kindest Regards,

Kelsey A. Jager

APPENDIX N

EXAMPLE OF INTEGRATED NARRATIVE FOR DESMOND

Appendix N

Example of Integrated Narrative for Desmond

Living with LGMD wasn't difficult for Desmond in the beginning. He did not see himself as different from his peers, just physically slower. The first symptoms of LGMD were trouble climbing stairs, which became noticeable when his family moved from a ranch to a 2-story home. His pediatrician referred him to a specialist.

What he remembers most about the diagnostic process is his parents and medical staff holding him down every time he was stuck with a needle—a phobia at the time. He did not understand why the tests were being doing and when he was diagnosed, he did not understand what it meant to live with LGMD. He didn't "feel" the effects of it. Doctors explained to him that in the future, walking and lifting his arms would eventually get more difficult. He did not want to believe that LGMD would affect him in such a significant way; he brushed it off and believed it was something that might happen in the very distant future. He tried to forget that he had it altogether, and believe he would live a normal life—accomplishing milestones of emerging adulthood. He lived in denial that LGMD would progress and was optimistic about the future.

It was the summer before 7th grade when he experienced LGMD in a profound way when he realized he was no longer able to run. He remembers his body not working like it did the summer before. This was a turning point when he had to confront that something was "different" about him, and that his symptoms were progressing. He felt like less of a person, disappointment, and guilt. His father seemed angry and upset when they would play sports and his physical weakness was apparent. These reactions led him

to believe he was not meeting his father's expectations and he internalized shame. He would feel guilty and nervous if he could not do things, like drive an un-adapted vehicle, especially if his father seemed angry with him for LGMD symptoms inhibiting him to do something "normally."

He did his best to hide living with LGMD, especially at school. He was afraid he would not be accepted by his peers if they knew. If he was not walking upstairs, on an uneven surface, or getting up from a seated position, he looked "normal" and that's what he wanted others to believe he was. He would lie about why he was/was not doing something to cover up LGMD. He would come up with excuses to not do things if it might bring attention to his physical struggles.

Times he became very aware of living with LGMD are when he would fall, or have trouble getting up from a chair—which were embarrassing and anxiety-provoking. His peers would make negative comments and laugh. He buried those memories because they are painful and depressing.

As his condition progressed—and he noticed more trouble walking and getting up from chairs/toilet—his anxiety worsened. When his progression got to the point where walking was difficult and he had a lot of trouble getting up from a seated position, his anxiety worsened. He felt vulnerable and unsafe at school. He began feeling claustrophobic in the sense that he felt stuck wherever he was seated, and could not get up quickly or without others noticing his difference. He worried frequently about unexpected physical tasks like fire drills or needing to use the bathroom. He dreaded teachers calling him to the front of the class and feared not being able to get up while

everyone was watching. During this time, his anxiety was high and he was having panic attacks. His school attendance declined.

As LGMD worsened, he thought more about it and developed specific and calculated ways to continue to complete physical tasks that are challenging to him, like getting in and out of a car. Travelling on an airplane feels claustrophobic since he is without his wheelchair and moving/maneuvering to the restroom is impossible (independently). New situations sometimes end up with him having to rely on strangers to carry/lift him for transportation, toileting needs.

It was a knee injury from a fall at his grandmother's house (from a rug with no traction) that forced to use a wheelchair for a period of time. It was a physically and emotionally painful period of his life. He was angry and blamed himself for not being more careful. He felt like he was making progress in recovery when he fell and injured himself again and it set him back mentally. He felt devastated and abnormal, inferior and weak, especially when comparing himself to his unaffected sibling. It was difficult for him to push himself physically anymore when he felt the ultimate outcome of being disabled is already set. Up until this point, he thought he could maintain his strength.

He felt defeated using a wheelchair at first, as if he had given up the fight against LGMD; like a battle he had lost. However, it also relieved anxiety about falling in front of peers and not being able to get up.

Living with LGMD has had a huge impact on him mentally and emotionally. The progressive, degenerative, non-curable aspects of LGMD discourage him. He describes living with LGMD to be like hitting brick wall after brick wall, each one taking a toll on

him physically, mentally, and emotionally. Inside he feels depressed, negative and worthless, but tries to hide it and come across as positive. He has been diagnosed with anxiety and OCD, and takes medication (Prozac 50mg) to help with symptoms. It is difficult to mentally cope with LGMD and fight back against the negative thoughts regarding his perceived limited success.

At the time of the interview, he did not have anyone to talk to about his feelings, so he tries his best to repress distress. No one understands what he is going through. Over time it builds up and he gets more anxious. He describes it to be like being stuck in purgatory feeling lonely, hopeless and worthless. He tries to reason his way through it, but it is not enough.

He has tried talking to his parents, but decided not to lean on them for support. Even to this day, it is difficult for him to talk about his struggles of living with LGMD with his family. They misunderstand his hesitancy to go out as social disinterest. His parents did not bring much attention to LGMD growing up, and told him everything would be okay and that he would live a normal life. No one talked to him about his differences. he did not want to ask for help. He felt like doing so would disappoint them, or that they would think he was lazy or that he would be blamed for the progression. He felt invalidated by his parents when he perceived them to judge him for feeling anxious, as if it was a mental weakness. He finds himself justifying his feelings and actions to his parents.

He feels hopeless he will find intimacy and assumes no one would choose to be with him because of physical limitations and he avoids rejection. He feels like his life is

not going anywhere and that he will never achieve moving out of his mom's house, getting married and working full time. He worries he will die alone.

He spent many years in college, but low motivation and financial restrictions led him to drop out. He is hopeless he will ever be able to complete his degree. He feels lost at sea, or like a hamster on a wheel, unable to move forward in life. He is on disability. Jobs he is interested in have weight requirements, which discourage him from applying. While he likes the idea of work, he fears he is too disabled to maintain employment.

When he thinks about the future living with LGMD, he fears getting progressively worse and being bedridden. He is unsure how bad his symptoms will get. He actively stops himself from thinking about the future because when he does, he feels sad and scared.

He feels anxious, uncertain, and unmotivated to set goals and achieve them. Life with LGMD has taken such a toll on him that he feels hopeless life will improve. He focuses on getting by day by day

His social life is limited because he calculates how much energy an outing will cost with how much fun he expects to have, and then determines if it is worth the effort. When he goes to an unfamiliar place, he thinks ahead about accessibility and whether he would be able to use the restroom. He calls his future planning "mapping out an escape plan." Sometimes he restricts liquids to avoid using the toilet. He tries to avoid being surprised by something not being accessible. At times, it appears people see his wheelchair more than him.

PT, therapy, SSRI medication, and muscle relaxers have been treatments utilized since diagnosis. He was denied a stair lift for his home. He received counseling services at the age of 18 but stopped going because it was unhelpful. He did not go the MDA clinic as a child, and started taking vitamin supplements as an adult to prevent secondary complications (vitamin D for bone strength).

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