Examining parental utilization of and barriers to psychological interventions in the Duchenne Muscular Dystrophy community

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Submitted to the Graduate Faculty of the

Graduate School of Public Health in partial fulfillment

of the requirements for the degree of

Master of Science

University of Pittsburgh

2021

UNIVERSITY OF PITTSBURGH

GRADUATE SCHOOL OF PUBLIC HEALTH

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Abstract

Background: Duchenne Muscular Dystrophy (DMD) is an X-linked neuromuscular condition. Parents of individuals with DMD report experiencing anxiety and depression symptoms. Psychological interventions including psychotherapy, psychiatry, and support groups have shown to be effective, yet tend to be underutilized due to attitudinal and structural barriers.

Methods: 230 parents of individuals with DMD were anonymously surveyed to examine utilization and barriers to psychological interventions during the time of their child's diagnosis and as the condition has progressed over the years. The Public Health Questionairre-9 (PHQ-9) and Generalized Anxiety Disorder-7 (GAD-7) was utilized for mental health screening. Distribution occurred through advocacy groups who reached out to their members via social media and email. Results were analyzed quantitatively using descriptive statistics.

Results: Most participants did not utilize psychotherapy, psychiatry, and in-person or online support groups during the diagnosis (67.4%, 85.7%, 77.8%,78.3%) or disease progression (56.7%, 80%, 72.6%, 67%) stage. The top three barriers identified for not utilizing psychotherapy and psychiatry were "I felt that I did not need to", financial reasons, and time constraints. The top three barriers for in-person and online support groups were lack of support group availability, "I felt that I did not need to", and time constraints. Common qualitative barrier themes across all interventions included: being emotionally overwhelmed, other support resources, COVID-19 pandemic, and lack of resource information/availability. PHQ-9 screening revealed 94.78% and

91.63% of participants experienced varying degrees of depression symptoms with 42.6% and 23.26% who experienced moderate to severe depression during the diagnosis and disease progression stage, respectively. GAD-7 showed that 94.78% and 93.95% experienced varying degrees of anxiety and 58.26% and 34.41% had moderate to severe anxiety during the diagnosis and disease progression stage, respectively.

Conclusions: Psychological interventions are underutilized by parents of individuals with DMD, yet a majority experience anxiety and depression symptoms. Low perceived need and lack of support groups were identified as major barriers. Healthcare workers, such as genetic counselors, involved in this community should use family-centered care, implement mental health screenings, and increase conversations regarding psychological interventions when appropriate. Furthermore, these results have public health significance in improving access to psychological interventions.

Table of Contents

List of Tables

List of Figures

Preface

I would like to acknowledge those who came on this research journey with me: my entire committee, my family, and the Duchenne Muscular Dystrophy organizations. I'd like to give thanks to my committee for being incredibly patient and supportive. It was a long journey to the finish line, and I appreciate each person more than words can describe. Secondly, I'd like to thank my family, especially my mom, for believing in me and supporting me through my entire life. This research is dedicated to and inspired by her. As a parent of an individual with a neuromuscular condition she understands the importance of psychological interventions. Thirdly, thanks to all of the organizations that helped distribute this survey to their members. Without their help the response rate would not be as high as it is. Lastly, I would also like to also dedicate this research to the entire Muscular Dystrophy community. As an individual with a neuromuscular disorder, words cannot describe how important this community is to me. I hope this research lays the groundwork for future research. As a genetic counselor I hope to continue making a positive impact in the community both professionally and personally.

1.0 Introduction

Duchenne Muscular Dystrophy is a neuromuscular disorder that causes rapid progressive muscle deterioration throughout the body. Pathogenic variants in the *DMD* gene cause symptoms in early childhood and are inherited in an X-linked recessive manner. Diagnosis usually occurs around age 4 or 5 years. Signs and symptoms include high creatine phosphokinase, late onset walking, waddling while walking, enlarged calf muscles, and difficulty climbing, running, and getting up off the ground. Over time the muscles of the upper arms, shoulders, hips, and thighs will progressively become weaker. Those affected typically need to use a wheelchair by age 13, have signs of cardiomyopathy by age 18, and do not live past the third decade of life due to respiratory and cardiac complications.[1]

Numerous studies have been conducted describing the experiences of parents/guardians of individuals diagnosed with Duchenne Muscular Dystrophy. The most stressful periods for parents and families of individuals with Duchene Muscular Dystrophy are the time of diagnosis, loss of ambulation, adolescence, and end stages of the condition.[2] One study found that 57% of parents of children with Duchenne Muscular Dystrophy self-reported that they had poor psychological adjustment which resulted in 50% and 31% having depressive and anxiety symptoms, respectively.[3] Another study showed that parents of children with this condition are significantly more likely to go through a depressive episode and have more distress than a national control group, therefore, counseling on appropriate therapies is strongly encouraged.[4] Appropriate therapies can include group therapy, psychiatry, or psychotherapy.

Even though referrals to psychiatric interventions might occur, there can be barriers that prevent a parent/guardian from acting on a referral. A study conducted on the primary care patient population found that some perceived barriers to psychotherapy include cost, time constraints, transportation difficulties, childcare or caring for sick/disabled loved ones, discomfort talking about personal issues, concerns about being seen while upset, discussing personal issues with a stranger, and stigma. The same study also reported that 59.5% of participants stated that at least one of the barriers mentioned would make it difficult to attend and participate in psychotherapy.[5] These barriers could translate over to other psychological interventions such as group therapy and psychiatry.

While the current literature describes the stressors that parents face, there is a lack of literature describing whether parents utilize psychological interventions in order to cope with their child's diagnosis and the stressors of caring for a child with DMD. This study aims to examine whether or not parents of children with Duchenne Muscular Dystrophy have utilized psychological interventions over the course of their child's diagnosis/disease progress. Barriers to the psychological interventions will also be examined. The survey was developed in Qualtrics and distributed through partnering advocacy groups that include the Muscular Dystrophy Association, Parent Project DMD, social media, and the list-serve within the neuromuscular clinic at Children's Hospital of Pittsburgh. The results of this study will help genetic counselors and health care providers within the neuromuscular community address the psychological needs of parents of children with Duchenne Muscular Dystrophy.

This study aims to:

- Aim 1: Develop an online quantitative survey using Qualtrics with a target audience of parents or guardians of individuals diagnosed with Duchenne Muscular Dystrophy

- Aim 2: Utilize the survey, which will be distributed through Muscular Dystrophy advocacy organizations' publications, list serves, and social media, to assess the following:
    - Whether or not parents/guardians of individuals diagnosed with Duchenne Muscular Dystrophy have utilized psychological interventions, such as psychotherapy, psychiatry, or group therapy
    - Reasons or barriers explaining why parents/guardians of individuals with Duchenne Muscular Dystrophy chose not to utilize psychological interventions
- Aim 3: Assess the survey results to identify any barriers to utilizing psychosocial interventions in order for genetic counselors to better meet the psychological needs of parents/guardians of individuals with Duchenne Muscular Dystrophy or other neuromuscular conditions.

2.0 Literature Review

2.1 Duchenne Muscular Dystrophy

2.1.1 Overview

Duchenne Muscular Dystrophy (DMD) is a neuromuscular condition that typically affects about on average 7 (range 1-16) per 100,000 males worldwide.[6-27] Although it is rare, it is one of the more common muscular dystrophies. It is caused by variants in the *DMD* gene, located on the X chromosome.[28] There are a variety of pathogenic variants that consist of deletions, duplications, and point mutations. These can cause absent or decreased production of dystrophin; an essential protein needed to form and maintain healthy muscle. Variants in the same gene can cause another type of Muscular Dystrophy called Becker Muscular Dystrophy (BMD). The difference between DMD and BMD is that out-of-frame deletions/duplications cause DMD while in-frame deletions/duplications cause the less severe BMD.[29] DMD is inherited in an X-linked recessive manner where female carriers of a pathogenic DMD gene variant have a 25% chance of having a child with DMD[28]. Female carriers have been reported to have some cardiovascular symptoms; therefore, it is recommended that they seek a cardiology evaluation.[30]

Individuals with DMD present with symptoms before age 5 years with mean age of diagnosis being around 3 years.[31-33] Initial symptoms include delayed motor milestones, difficulty climbing stairs, waddling, persistent toe walking, elevated creatine phosphokinase, calf hypertrophy, and the classic Gower maneuver.[33-37] As the disease progresses there is symmetric muscle weakness with the proximal muscles being more affected than the distal muscles. By age

13 years, most individuals utilize a wheelchair full time for mobility needs due to loss of ambulation.[38] The condition not only affects the skeletal muscle, but also cardiac and respiratory muscle. During the later stages of DMD, individuals may require ventilation support due to breathing difficulty. Involvement of cardiac muscle leads to dilated cardiomyopathy resulting in heart failure. Cardiopulmonary complications are usually the cause of death within the second to third decade of life.[39] However, life expectancy has increased over the years as new treatments emerge.[39-43] There is no known cure for DMD. Treatments and management guidelines are strictly supportive but can delay progression. Treatments such as exon-skipping and stop-codon-read-through therapy are available depending on the individual's genetic variant and stage of disease. Gene therapy is currently being studied at the research level.[44-45]

2.1.2 Molecular Genetics

Duchenne Muscular Dystrophy is caused by variants in the *DMD* gene located on chromosome Xp21.2-p21.1. It is the largest known gene encompassing 79 exons.[46] It is expressed mainly in skeletal muscle, cardiac muscle, and at low levels in the brain.[49-50] It is not found in any non-muscle tissue. The *DMD* gene is responsible for making the dystrophin protein. Dystrophin is a rod-like structure located in the inner surface of muscle fibers, called the sarcolemma.[48] The protein has four main functional domains: actin-binding amino-terminal domain, central rod domain, cysteine-rich domain, and carboxyl-terminus domain.[47] Dystrophin acts as an anchor between the dystrophin-glycoprotein complex embedded along the sarcolemma and intracellular actin network. This linkage is critical for muscle stability.

There is a wide spectrum of variants that cause DMD. In the past, variants in the *DMD* gene were hard to detect due to how large the gene is. Approximately 66% of variants are large

(one or more exons) deletions and about 5% are large duplications.[29,51-53] More advanced genetic studies have identified point mutations (10-30%) and splice site variants (2%) to the spectrum of variants.[54-58] Recently, a nationwide study in Italy involving 11 diagnostic centers genotyped 1,902 patients over a 10-year period and found that in DMD patients the spectrum of variants was deletions (57%), duplications (11%), and point mutations (32%), 44% of which were nonsense mutations.[59]

2.1.3 Inheritance

Duchenne Muscular Dystrophy is inherited in an X-linked recessive manner. The *DMD* gene is located on the X chromosome. Males are primarily affected. About 30-33% of the time the condition occurs from a *de novo* variant meaning that the variant was new in the child and not inherited from the mother.[60-62] According to Haldane's law, a female who has one son with DMD has a 67% chance of being a carrier. This risk can be lowered depending on any unaffected sons present. A female with two sons with DMD is considered an obligate carrier. Female carriers are typically asymptomatic. When a carrier female becomes pregnant there is a 25% chance of having a son with DMD, a 25% chance of having a son without DMD, a 25% chance of having an unaffected carrier daughter, and a 25% chance of having an unaffected daughter who is not a carrier. Although carrier females are typically unaffected, some may experience symptoms of dilated cardiomyopathy, muscle weakness, and muscle pain/cramping with variable expressivity.[63] These symptomatic females occur in 8% to 22% of DMD carriers.[64-65] In more rare cases female carriers can present with DMD symptoms or a milder phenotype and are classified as symptomatic or manifesting carriers.[66-68] Studies have shown that this situation in females who are heterozygous

for dystrophin mutations can occur due to causes such as skewed X-inactivation and a chromosomal translocation involving the X chromosome.[67,69-72]

2.1.4 Diagnosis

When it is suspected based on history and physical exam, there are numerous ways to establish a diagnosis of Duchenne Muscular Dystrophy. Diagnosis typically occurs around 5 years of age.[73] The investigative techniques are muscle biopsy, serum creatine kinase, and genetic testing. In the past when genetic testing was not as prevalent or advanced as it is today, a muscle biopsy was a prominent technique to aide in the diagnosis.[74-75] The level of dystrophin using Western blot analysis will either be very low (less than 3% of what is considered normal) or completely absent.[76-77] When there is a lack of dystrophin there is muscle fiber degeneration and necrosis which gives rise to smaller cells as replacement.[79-82] Additionally, inflammatory cells can be present in response to necrosis.[83] Overtime the muscle degeneration surpasses the regeneration capacity which results in increased level of connective tissue and fat giving the appearance of pseudohypertrophy followed by atrophy.[78] More recently this practice is not utilized unless genetic testing cannot confirm a diagnosis.

The second investigative approach when DMD is suspected is to obtain a serum creatine kinase (CK) level. Creatine kinase is an enzyme that is commonly found in the heart, skeletal muscle, and brain. In unaffected individuals the CK level ranges from 39-308 U/L for males and 26-192 U/L for females.[84] Individuals with DMD can have an elevated CK level that is 10-200 times the reference value before the age of 5 years.[85] This approach has gradually replaced the need for a muscle biopsy due to the approach being less invasive.

Molecular genetic testing is the gold standard for a diagnosis of DMD. In the past multiplex polymerase chain reaction (PCR) was the main method used to identify variants because the majority of individuals with DMD had deletions of one or more exons.[86-87] Multiplex PCR was able to detect 98% of deletions, but it is unable to detect duplications or point mutations.[87] Multiplex ligation-dependent probe amplification (MLPA) or comparative genomic hybridization array can identify both deletions and duplications, even small ones, with the possibility of predicting if the deletion or duplication will disrupt the reading frame.[88-90] MLPA has the ability to improve the detection rate of multiplex PCR by 15%.[88] Oligonucleotide-based array comparative genomic hybridization (array-CGH) is able to detect complex rearrangements and large scale intronic alterations.[91] If deletion or duplication analysis comes back negative, then next generation sequencing should be performed to identify any point variants[92]. A study conducted in 2011 showed that next-generation sequencing was able to identify point variants, mainly nonsense or frameshift variants that caused truncation of the dystrophin protein, in 15 out of 16 (93%) participants who were not found to have a deletion or duplication.[93] Overall, molecular diagnostic methods for DMD have 90.7% sensitivity, 66.4% specificity, 93.2% positive predictive value, and 58.5% negative predictive value.[94]

2.1.5 Natural History

The natural history of Duchenne Muscular Dystrophy (DMD) can be broken up into five major stages: diagnosis, early ambulatory, late ambulatory, early non-ambulatory, and late non-ambulatory. It is important to keep in mind that the onset and duration of each stage can vary between individuals and can be influenced by medical and nonmedical interventions. The diagnosis stage typically occurs around 2 to 5 years of age.[31-33] Some of the most common

symptoms during the diagnosis stage include gross-motor delay (42%), delay in walking (mean age 18.3 months) (20%), toe-walking and flat footedness (30%), and less commonly learning difficulties (5%) and speech delay (3%).[33-34] Additional symptoms include difficulty running, climbing stairs, jumping, and standing up.[35] The disease course begins in the proximal lower limb muscles then affects the upper distal limb muscles as the disease progresses over the years.

When the individual is between 3 to 6 years old, it is known as the early ambulatory phase. This is where there is a waddling gait, lumbar lordosis, calf pseudohypertrophy, calf pain, and the Gowers' sign. The calf pseudohypertrophy is due to the muscle fibers being replaced by fat and connective tissue.[37] The Gowers' sign involves an individual utilizing hand to floor support with their legs spread apart and then crawling up their thighs with their hands for support to achieve a standing position, which occurs due to the weakened pelvic girdle muscles.[36] In addition, preclinical cardiac symptoms can occur. Pre-clinical cardiac involvement is seen in 25% of individuals under age 6 years and 59% between ages 6-10 years[102]. James et al. 2010 examined the prevalence of electrocardiography abnormalities in children with DMD under 6 years old in order to assess correlations between electrocardiography and echocardiography evidence of cardiomyopathy.[98] As a result, 78% of individuals were found to have electrocardiography abnormalities however only one echocardiogram was abnormal. This study concludes that electrocardiography abnormalities are quite prevalent in early stages of the condition well before the clinical onset of cardiac symptoms.

Between the ages of 6 to 11 years, the late ambulatory stage, there is rapid muscle deterioration over a 2–3-year period where the individual can quickly lose the ability to climb stairs even with rails, achieve a standing position, or walk a short distance (750 cm).[95] Joint contractures occur at the ankle, knee, wrist, elbow, and hip which inhibit mobility. The ankle

contractures lead to persistent toe walking[96]. Individuals typically use leg braces around age 10 years to ambulate.[37] In addition, beginning before age 10 years deep tendon reflexes are starting to be difficult to elicit in the knee, tricep, and bicep (50%). The ankle reflex could be elicited in 33% of individuals even during the final stages of the condition.[97]

Between the ages of 11-13 years old, the early non-ambulatory phase, weakness in upper and lower extremities progresses. A wheelchair is the main mode of mobility as this is the age range where loss of ambulation typically occurs.[38] Scoliosis begins at the average age of 13.29 years due to muscles of the trunk becoming weaker.[31,100] Respiratory muscles become weaker thus beginning the decline in respiratory function.[101] During the late non-ambulatory phase, which occurs in the late teens to late 20s, respiratory failure in addition to cardiac failure, due to dilated cardiomyopathy, starts to occur. Respiratory failure is the leading cause of death at an average age of 17.7 years.[39] Dilated cardiomyopathy is the sole cause of death in only 20% of individuals with DMD at the average age of 19.6 years.[39,103] Nigro et al., 1990 found that clinical cardiomyopathy is typically evident around 10 years old and is seen in 33% of individuals by age 14, 50% by age 18, and 100% of individuals over age 18 years.[102] Typically, median survival is around 19 years, but there has been an improvement in survival. A study conducted in 2012 found a significant improvement in survival where those born more recently (i.e., 1980-1989) had a higher chance of surviving beyond age 20-25 years.[39] This is due to the advancements in management and treatments.[40-42] Another study found that there was an 85% probability to survive to age 30 years.[43] Even with medical advancements individuals typically do not survive beyond the third decade.[39,43]

Other features of DMD include neurocognitive, gastrointestinal, and decreased bone health. Cognitive features include learning difficulties such as difficulties in verbal and reading skills, and verbal memory, though the extent is variable and there are other factors such as physical

disability, environmental factors, and age that could influence these features.[104-110] Banihani et al. 2015 and Ricotti et al. 2016, found that attention-deficit hyperactivity disorder (ADHD), autism spectrum disorder (ASD), learning disabilities, and anxiety were present in 32%, 15%, 44%, and 27% of individuals with DMD.[111-112] Several studies have found that as an individual's condition progresses, they may experience depression or anxiety symptoms.[113-115] It is imperative to counsel individuals on and be sure they have appropriate support systems that can aide in minimizing these symptoms. Other behavioral aspects include lack of attention span, executive control difficulties, as well as poor social skills.[116-119] Gastrointestinal features are quite common in individuals with DMD. About 8 out of 11 individuals can have a range of symptoms including but not limited to delayed gastric emptying, acute gastroparesis, and abdominal pain.[120-121] Lastly, individuals with DMD are at significantly increased risk for osteoporosis when they lose ambulation, and it is more severe in the lower limbs.[122] This can cause an increased risk for fractures.

2.1.6 Treatment and Management

Management for individuals with Duchenne Muscular Dystrophy is comprehensive and multidisciplinary due to the involvement of multiple systems. Similar to the natural history of the condition treatment and management protocols for DMD are implemented in five stages: diagnosis, early ambulatory, late ambulatory, early non-ambulatory, and late non-ambulatory. Since features of the condition can present in slight variation of age between individuals, the implementation of care should occur on a continuum. A neurologist who specializes in neuromuscular diseases, typically at a neuromuscular care center, should lead the multidisciplinary team and assess the patient every 6 months following diagnosis. This doctor is responsible for assessing the individual's strength, function, and range of movement as well as advising on new

therapies, educating patients and their families, and providing support. During the diagnosis and early ambulatory stage, discussion, initiation, and management of glucocorticoid steroid treatment should occur. Corticosteroid therapy significantly improves strength, lung function, motor function, and delays onset and progression of cardiomyopathy.[123-125] Balaban et al. 2005 examined the long-term functional status in males with DMD who were taking prednisone, deflazacort, or no steroid use.[124] It was discovered that males taking either steroid brand significantly retain function and have slower disease progression than males in the control group. Both steroid brands were equally effective. Steroid therapy also helps prolong ambulation and delay scoliosis because of the prolonged ambulation.[126-127] Takeuchi et al. 2013 discovered that the age at which individuals with DMD lost ambulation was significantly older in those who took prednisone compared to those who did not.[126] Side effects of corticosteroids include excessive weight gain, short stature, facial fullness, behavioral changes, gastrointestinal complications, blood pressure changes, hypertrichosis, acne, cataracts, and decreased bone health.[124,128-131] The challenge with glucocorticoid steroids is what dosage provides the greatest benefit while minimizing the side effects.[132] This challenge can result in variations of undertreatment or overtreatment. Other specialists that are important to the management of DMD include Rehabilitation, Cardiologist, Pulmonologist, Orthopedist, Neuropsychologist, Endocrinologist, and Gastroenterologist or Nutritionist.

Rehabilitation management includes providing referrals for occupational, physical, and speech therapy to maintain mobility, conserve energy, prevent injuries, manage pain, and learning support. They also coordinate the provision of mobility devices, standing devices, and assistive technologies. Cardiology will implement cardiac function management. During the diagnosis stage a baseline electrocardiogram and echocardiogram should occur. Cardiac function should be

checked annually or sooner if symptoms are present. Angiotensin-converting enzyme (ACE) inhibitors are a first-line management choice for those with dilated cardiomyopathy. Individuals with DMD should be placed on this medication by age 10 years.[145] Duboc et al., 2007 examined the ACE inhibitor Perindopril's preventative effect on mortality in males with DMD whose left ventricular function was within normal limits over a 10-year period.[146] The results indicate that the medicine was able to significantly lower mortality. Pulmonology should be involved to assess lung function every 6 months starting in the early ambulatory stage. Pulmonary function tests should begin around age 8 or 9 years.[101,147] Nocturnal and daytime ventilation and cough assist can be initiated at the end of the early non-ambulatory stage when lung function starts to decrease.[145] Orthopedics manage contractures, range of motion, and scoliosis. Spinal fusion can be considered in certain circumstances during the early non-ambulatory to late non-ambulatory stage. Individuals who lose ambulation later were less likely to require spinal surgery. Those who had spinal surgery and nocturnal ventilation have a median survival of 30 years, and those only using nocturnal ventilation had a median survival of 22.2 years.[41,148] Neuropsychologists can evaluate and provide resources for any learning, emotional, or behavioral concerns. Nutritionists and gastroenterologists can aide in maintaining a healthy weight, vitamin D and calcium levels, swallowing function, and minimizing gastric upset.

Treatments such as exon skipping therapy and Ataluren are available for individuals with certain DMD gene variants. Exon skipping, where the cell's transcriptional machinery is made to skip an exon containing a deletion or duplication that would cause DMD, allows DMD to be converted to the milder BMD phenotype.[133-134] In theory 83% of individuals can be treated with this technique.[135] Exon skipping therapy for exons 51,[136-137] 45,[140] and 53[139] are available and deemed safe and preliminarily effective.[138] Studies show that the exon skipping therapy causes a

significant increase in dystrophin and sustained ambulation compared to controls. This type of therapy is only available for individuals with deletions of specific exons which results in ~29% of the Duchenne population being eligible to receive the treatment.[135]

Ataluren is available for the 11% of individuals with point mutations, specifically nonsense variants, within the DMD gene. Nonsense variants cause a premature stop codon to be created, thus disrupting transcription. Ataluren acts to ignore the stop codon and continue to make the dystrophin protein.[142] Studies showed that the treatment allowed for a slower decline rate in the six-minute walking distance measure than the placebo group, though the difference was found not to be statistically significant.[141] Gene therapy is currently being utilized at the research level.[44-45] As gene therapy is being developed, scientists and clinicians will have to address patients having dystrophin-specific T-cell immunity which could affect the success of the treatment[143]. This risk has been shown to increase with age, but glucocorticoid steroid treatment can decrease the risk.[144]

2.2 Parental Experience

2.2.1 Stages of Grief

When parents learn that their child has a disability or chronic illness, they experience a grief response that is similar to that of when a child dies[153-155]. Adjusting to the fact that their child is different from what they were expecting, parents often go through multiple stages of grief developed by Kubler-Ross. The five stages of grief are denial, bargaining, depression, anger, and acceptance.[149] The stage of guilt can sometimes be substituted for bargaining.[156] These stages should not be thought of as a linear process. There is no guidebook or timetable for how the

grieving process should unfold. The process is extremely individualistic and has great variety between individuals. Kubler-Ross 1969 based her grief model on observations of the terminally ill. [149] While this model fits with the end stages of DMD, other grief models have been developed that may be applicable to families during the diagnosis and early stages of DMD. Therese Rando developed a mourning process model consisting of the "Six R's": recognize, react, recollect and/or re-experience, relinquish, readjust, and reinvest[150]. Rando's model can be applied to how parents cope and adjust to their child's diagnosis. Margret Stroebe and Henk Strut's "Dual Process" model of grieving describes how an individual deals with loss by oscillating between the internal loss orientation and restoration orientation. The internal loss state is where the individual focuses on the loss that they are experiencing and the motions that surround it while the restoration state focuses on aspects of life that we need or want.[151] As a result, the loss is processed over time in the capacity at which the individual is capable of while dealing with aspects of everyday life. Lastly, Schneider 1983 provides a holistic approach to grief that not only examines how grief affects an individual in the biological, emotional, and behavioral sense, but also one's intellect, spirituality, and attitude.[152] The model promotes growth and self-awareness. In Schneider's model there are six stages of grief: initial awareness, strategies to overcome loss, awareness of loss, completions, resolution and reformulation, and transcending loss.[152] At first each new roadblock may trigger feelings of frustration but overtime the coping tools become stronger. By the time the individual is transcending the loss they are no longer inhibited by the emotional weight of it. They can use their newly found energy to seek activities that would bring enjoyment to their child, themselves, and the family.[152,157] These models may help us to understand what parents are going through but will not be reflective of all experiences.

2.2.2 Children with disabilities or chronic illness

Coping is the constant changing of both cognitive and behavior efforts to manage a situation, whether it be internal or external, that is beyond the person's capability at that specific moment in time.[158] As discussed above coping is an integral part of the grieving process; therefore, coping itself is a continuously changing process that is rooted in the context of the situation. Strategies for coping constantly evolve as the root of the stress unfolds. It is no surprise that parents are a significant part of their child's life, growth and development.[171] When a child with disabilities is born, parents not only have to cope with the same parental stressors as the general population, but parents must cope and adapt to stressors unique to the child's disability.[173-175] Some studies concluded that parents of children with chronic illness adapt no differently than parents who do not have a child with chronic illness.[176-178,181] Breslau et al., 1986 found that there was no significant difference in the rate of major depressive disorder between mothers of children with disabilities and the control sample.[177] Kovacs et al., 1985 revealed that parents were mildly or subclinically depressed or anxious during their child's diagnosis, then after 6 months of going through the grief process the symptoms subsided.[178] On the other hand, there have also been numerous studies supporting greater levels of psychological stress in parents who have children with a disability or chronic illness compared to controls.[160,180] The fact that there is a large variety of chronic illnesses that vary in severity along with variation between parents' attitudes, beliefs, and abilities might account for lack of cohesiveness in the literature.

There are specific time periods where parental stress occurs. Clements et al. 1990 investigated parental experiences that were deemed difficult in regard to caring for a child with a chronic illness.[159] The diagnosis period and times of disease progression were found to have high amounts of stress. At diagnosis parents felt guilt, shock, hopelessness, uncertainty, isolation,

denial, fear, anger, confusion, and depression.[164-165,169-170] Due to these psychosocial stressors, parents are significantly more likely to report psychological symptoms with mothers reporting more often than fathers.[163-164,166] In addition to the parental experience at diagnosis Heiman 2002, explored parents' experience after the diagnosis period had completed. The author found that the majority of parents reported never-ending emotional fatigue, social isolation, lack of freedom, and had an unmet need for information on social and psychological resources. In the same study 75% of parents felt that the feelings they experienced at the diagnosis stage had turned into joy, love, happiness, and satisfaction. On the other hand, 25% still felt anger, guilt, sadness, and frustration.[165] The fact that some parents still felt sadness and frustration correlates with the concept of chronic sorrow. This term described by Olshansky 1962 states that while grieving can vary, in regard to intensity and time, it continually affects the individual.[162] It is often described in a wave-like fashion. This data endorses the fact that the parental experience, while noting similarities, is individualistic.

Numerous studies have investigated how parents of a child with a chronic illness or disability cope. One study found that maintaining family cohesion, intellectualization, and maintaining social support aided in parental adjustment.[172] Other studies showed similar coping mechanisms with the addition of using direct efforts such as planning, taking control, and problem solving as well as using different approaches to life such as hope, living in the moment, and not dwelling on difficulties.[161,168] Parents tend to significantly use more avoidant coping, lowered belief that life situations will work out as well as expected and have less focus on personal growth compared to parents who do not have a child with a disability.[179] Avoidant coping can be recognized in parents who focus on the needs of their child with disabilities above all else.[170] Both mothers and fathers have the capability to adapt and have close-knit social support networks.[166-

[167] Clements et al. 1990 reported that when parents have resources to attend to the emotional and physical aspects of caring for a child who is chronically ill, distress symptoms are minimized.[159]

2.2.3 Children with Duchenne Muscular Dystrophy

By nature, Duchenne Muscular Dystrophy falls under the category of disability and chronic illness. This condition, for which there is no cure, causes progressive muscle weakness throughout the body resulting in a physical disability. Due to this it is reasonable to see similarities and differences between coping for parents of children with DMD, other chronic illness/disability, or no chronic illness/disability. Parents of children with Duchenne Muscular dystrophy display a similar experience, if not a more significant level of psychological stress, to parents of children with other chronic illnesses.[190] They also experience more stress, five times more, than parents of children without a chronic illness/disability.[191-194,198] Miller, 1990 demonstrated that the most stressful periods for parents and families of individuals with Duchene Muscular Dystrophy are the time of diagnosis, loss of ambulation, adolescence, and end stages of the condition.[2] At the time of diagnosis parents feel angry, sad, depressed, low self-esteem, fear, guilt, confusion, powerlessness, overwhelmed, uncertain, anxiety, anguish, and shock.[4,186,199-201] Of course, these feelings are similar to those grounded in grief models. Some parents want psychosocial support to be readily available while others are simply not ready.[3,183,201]

As their child's condition progresses some parents still feel anxiety, overwhelmed, uncertainty, low self-esteem, and/or depressed.[4,185-186] They experience chronic sorrow where there is a continuous period of loss, and adaptation that challenges their coping ability each time as the condition progresses.[182,188,201] In the study conducted by Saetrang et al. 2019, one parent even described how the sorrow comes on suddenly causing immense exhaustion and feelings of being

alone in their grief.[201] This parent even recognized that both the child and them should receive professional help to deal with the life-limiting aspect of the condition as it progresses, but they did not feel that they were in a position to bring up the concerns during their child's medical appointment. Reid et al., 2001 and Gocheva et al., 2019 found that the level of family stress significantly predicts the psychosocial adjustment of the child with DMD.[193,198] These findings provide further evidence on the efficacy of treating the family in a holistic manner or even introducing palliative care at some point for emotional and spiritual support.

Often parents are the sole caregiver for their child throughout the child's lifetime (78.1%).[189] Numerous studies have shown that overall parents are at an increased risk for developing or experiencing depression or anxiety.[3-4,185-187,195-197] 31-80% of parents report experiencing moderate or severe depression or cried sometimes, often, or always[3-4,186-187]. As for anxiety, 21-50% report experiencing moderate to severe anxiety where there is constant worrying.[3,185,187] In addition, 50-60% of parents report poor sleep quality, reduced sleep efficiency, and daytime dysfunction, which can exacerbate mental health symptoms.[185-186,196,201-204] Due to the prevalence of psychological symptoms and distress it is recommended that parents receive counseling on appropriate psychological interventions.

Several studies have investigated how parents of children with DMD cope and adjust. Some utilize maladaptive coping styles, such as magical thinking, overprotection, internalization, and passive coping.[3,184,193,201] Those who use these coping methods tend to have higher distress. Passive coping can occur when the psyche deems the situation too hard to talk about and has shown to have a correlation with anxiety where those who use this coping strategy have higher levels of anxiety.[3,185,201] Since these maladaptive coping styles occur, psychological intervention could be used to introduce better coping strategies to minimize distress. Studies have shown that parents

want support resources to develop better coping skills and parents have the ability to cope well if given the appropriate resources.[183,199,201] Saetrang et al., 2019 found that parents needed professional help to work through the shock of their child's diagnosis.[201] Support resources can encourage coping strategies that parents describe as successful in minimizing distress such as living in the moment and appreciating present abilities, intellectualization, being proactive, and setting short-term goals.[183,199,201] Mah et al., 2012 states that the need for psychological support may decrease slightly as the disease progresses due to better coping strategies, adjustment, and an accepted reality.[183]

2.3 Psychological Interventions

2.3.1 Psychotherapy

According to the American Psychiatric Association psychotherapy is talk therapy with the goal of aiding individuals with mental health conditions or emotional difficulties minimize symptoms to increase healing and functionality.[205] Dwight-Johnson et al. 2000 examined treatment preferences among individuals with depression. 83% of participants wanted some form of treatment with 67% preferring psychotherapy.[206] Psychotherapy has also been shown to be more cost-effective than psychiatry.[207] The types of therapy, including cognitive behavioral therapy,[208-214] interpersonal therapy,[215-217] psychodynamic and psychoanalysis therapy,[219-223] and supportive therapy,[224-230] have shown to be effective in treating mental health conditions. Rush et al., 1977 found that psychotherapy showed significant improvement for patients with depression compared to pharmacotherapy. 78.9% of the patients in therapy showed marked improvement or complete

remission of symptoms compared to 22.7% of the pharmacotherapy group.[208] In addition, the dropout rate was significantly lower for the therapy group. Stanley et al. 2003, showed that psychotherapy was able to significantly reduce anxiety severity, worrying, and depressive symptoms.[214] Psychotherapy can also increase social functioning, problem-solving skills, and healthy coping mechanisms.[218] The different types of therapies have proven to be effective on their own, so researchers have compared the therapies on their effectiveness. Numerous studies have shown that there are no significant differences between the types of therapy and patients can equally benefit from each.[229-233] Any differences between therapies could simply depend on the patient's preference or their specific needs. This shows that patients psychotherapy treatment can be individualized for which method suits their needs and personality.

2.3.2 Psychiatry

Psychiatry is a form of medical care in which a medical doctor specializing in mental health examines the mental and physical aspects of mental health conditions to diagnose, treat, and prevent them.[234] The most common treatment that psychiatrists are involved in is the prescription and management of psychiatric medication such as anti-depressants, anti-psychotics,[235-236] stimulants,[237-239] and anxiolytics. Anti-depressants are used to treat depression,[215,249-250] anxiety,[241,244] panic disorders,[240,243,245-246] post-traumatic stress disorder,[242,247] and obsessive-compulsive disorder.[248] An international collaborative study found that antidepressants were prescribed in 7.7% of anxiety conditions compared to 31.9% of depressive conditions.[251] Malt et al., 1999 found that antidepressants were effective in treating depression, even recurring depression, compared to a placebo control group.[250] Although there are countless studies confirming the effectiveness of antidepressants, about 50-55% of patients with depression are

treatment resistant, meaning they do not respond to the medication.[252-253] Treatment resistance has been correlated with other psychological and medical comorbidities. This includes anxiety, personality, and bipolar disorder and heart disease, cancer, and diabetes.[254-257] Those with treatment resistant depression are also at increased risk of morbidity, high medical costs, and lower quality of life.[258-263] Anxiolytics are utilized to treat anxiety conditions. An example of an anxiolytic are benzodiazepines. While benzodiazepines are highly effective and relatively safe when used for acute anxiety conditions,[264-268] they are not ideal for chronic generalized anxiety disorder (GAD).

2.3.3 Support Groups

Support groups by definition are comprised of a group of individuals who share a similar life stressor, transition, or affliction engaging in mutual support to improve coping and adjustment, alleviate loneliness, facilitate personal empowerment, and offer a sense of community.[271,275,301] This is done through listening to and sharing information, feelings, various coping strategies, and personal experiences. 40% of Americans had been a member of a supportive group at some point in their lives.[272] Social support is imperative for families of children with chronic illness or disabilities.[273-274] Parents report that they do not obtain the same level of support from family friends or health care professionals compared with support groups.[169] Studies looking at the efficacy of support groups showed that the intervention was able to significantly reduce anxiety, stress, depression, and the risk for mental health illness.[275,277-280] Social support increased feelings of coping, self-confidence, and optimism while significantly decreasing feelings of helplessness.[278] Another study found that those who do not participate in support groups have significantly higher levels of depression, anxiety, phobic

anxiety, feeling personal inadequacy/inferiority compared to others, and paranoid ideation.[282] Parental experience in support groups was studied for parents within the Duchenne Muscular Dystrophy community. Kornfeld et al., 1979 found that parents were able to express fears or concerns, learn from each other, and increase their awareness of attitudes toward themselves, their children (with or without DMD), and the world in general, which helps facilitate coping.[188]

A common example of a support groups for parents of children with chronic illness or disabilities is the Parent-to-Parent network. Parents of children with disabilities are uniquely qualified to help each other since they possess the knowledge of the 24-hour-a-day reality of their child's condition that others not in that situation do not possess. Parent-to-parent network was able to show a statistically significant gain in accepting family and disability (adaptation). 89% of parents rated the parent-to-parent group as helpful.[276] The parent-to-parent network is intended to connect parents who have a child with the same condition. This sense of sameness between the parents is crucial and there is support theory that supports this.[285-288] Thoitis 1986 and Taylor et al., 1990 describe how when there is perceived sameness within support groups it makes the advice and information shared have an increased level of credibility.[285,288] A successful parent-to-parent match is when there are not only similar child circumstances but also parental personalities and background. Parents report that they gained a realization that they were not alone, found individuals who truly understand their lived experience, were able to offer support to others, gained confidence and a sense of normalcy, and learned through social comparison from parents whose children were older.[169,270] With social comparison there can be positive upward comparison that gave hope or positive downward comparison where parents counted their blessings that their child was not worse off compared to other children.[290] In addition, parents were able to quickly reach a stage, a few years after their child was born, where the need for support lessened. Even though

they reach this stage, contacts and friendship were sustained. However, attendance to meeting tended to decrease as the child's age increased.

Those who are dissatisfied with offline support are significantly more likely to get support online.[269] There's a sense of anonymity that puts one at ease especially with the fear of stigmatization.[301] Having the support group online is a cost saving for society while providing support for those who were experiencing an emotional toll, needing a place for catharsis, seeking specific advice, or to problem solve.[289] Oprescu et al., 2013 found that information discussed or shared is primarily personal experience (87%) rather than medical information (13%) to manage uncertainty.[300] They also found that women tend to utilize online support groups more than men.

With all the positive aspects of support groups there can also be negative aspects. Often times in support groups information regarding the specific health condition can be shared.[281] Due to this there is a potential for misinformation to be spread. One study found that information on a bariatric surgery Facebook group was 7% inaccurate and 29% may or may not have been inaccurate but needed more context to determine accuracy.[284] On the other hand, another study found that information on a breast cancer support group site was false or misleading 0.22% of the time and of that 70% were corrected for accuracy.[283] It is important that the source and content of the information being spread is validated. Social comparison produces effective support because experiences are validated. However, social comparison can turn negative when differences between parents arise and can further exacerbate isolation, feelings of inferiority, and distress.[285] The differences can include different communication styles, outlooks on disability, and differing beliefs and parental styles.[270] There can be negative upward comparison where parents feel inferior compared to others and negative downward comparison where parents see other children at a more advanced stage of the disease felt distressed because they are confronted with the realization that

eventually their child will reach that stage. When negative comparisons occur, it is important for parents to reflect on the common ground that brings the community together, engage with another parent that has a higher level of sameness, or avoid social comparisons by not attending a support group.[290]

2.4 Barriers to Psychological Interventions

2.4.1 Common Barriers

Studies conducted in the United States of America as well as across the world show that a majority of individuals experiencing mental health concerns remain untreated or do not seek treatment.[311,313-315] Blumenthal et al., 1996 reports that 45% of individuals seek treatment for their mental health concerns while 56% do not.[315] They also found that those who had sought treatment in the past were significantly more likely to seek treatment again. Other studies found that approximately 20% of individuals referred to psychotherapy go through with utilizing the psychological intervention.[316-317] Since most individuals do not utilize psychological treatments, it is reasonable to question what barriers or reasons prevent individuals from seeking treatment. Typically, the types of barriers can be divided into two categories: attitudinal and structural.

Attitudinal barriers are internal factors or reasons that affects utilization of psychological interventions. Numerous studies have shown that internal beliefs and attitudes affect a person's receptivity to psychological interventions.[315,318-321] Pyne et al., 2005 concluded that those who have negative attitudes towards psychiatric medications are less likely to be prescribed the medications, less likely to fill the prescription, and less likely to achieve beneficial outcomes.[321] Jorm et al.,

2008 studied this occurrence further and discovered that those who have negative attitudes about psychiatry tend to be equally or more negative about other psychological interventions and may reject psychological treatment altogether.[320] Other attitudinal barriers include believing in handling the mental health concerns alone, low perceived need or severity of concerns, believing treatments would not help, and stigma.[310,315] Furthermore, mental health symptoms, such as depression, can further exacerbate attitudinal barriers such as lack of motivation, emotional concerns, negative feelings toward therapy, and stigma.[5]

Structural barriers are external reasons that prevent an individual from seeking psychological interventions or having access to psychological interventions. A study conducted on the primary care patient population found that some perceived barriers to psychotherapy include cost, time constraints, transportation difficulties, and childcare or caring for sick/disabled loved ones. The same study also reported that 59.5% of participants stated that at least one of the barriers mentioned would make it difficult to attend and participate in psychotherapy.[5] In the past, numerous studies found that financial costs tend to be a major structural barrier due to high costs or lack of insurance coverage.[306-309] Wang et al., 2005 found that those in low-income, low education, urban areas have higher treatment inadequacy.[311] However, the implementation of Health Maintenance Organizations (HMOs) and the Affordable Care Act, individuals have better access to psychological interventions.[302-305] Lastly, for parents of children with a disability or chronic illness, the main structural barrier tends to be the lack of time.[270] Understanding the prevalence of these barriers allows for health care providers as well as public health workers to create ways in which these barriers can be minimized to improve access to psychological interventions especially when there is a clear unmet need for a group of individuals.

2.5 Summary

Overall, there is countless literature describing the experiences and feelings of parents of children with various chronic illnesses or disabilities. Parents of children with Duchenne Muscular Dystrophy are no different. Research has been conducted describing their experiences during their child's diagnosis, how they felt, the stages of grief that they go through, the stress they continue to feel as the disease progresses and the grief of losing their child to this life-limiting condition. It is known, without a doubt, that parents within the DMD community can experience mental health symptoms such as anxiety and depression. However, there seems to be a lack of literature describing the proportion of parents who utilize psychological interventions such as psychotherapy, psychiatry, and support groups. There have been qualitative studies showing that parents of children with DMD find support groups helpful, but as for the other interventions there are no studies to our knowledge. For this reason, this study will aim to capture the utilization rates for each psychological intervention. In addition, this study will examine the barriers to these interventions. Many studies have described barriers to each intervention, but to our knowledge no research has been conducted to examine barriers to psychological interventions for parents within the DMD community.

3.0 Manuscript

3.1 Background

Duchenne Muscular Dystrophy (DMD) is an X-linked neuromuscular condition that typically affects on average 7 (range 1-16) per 100,000 males worldwide.[6-27,28] It is caused by variants in the *DMD* gene, located on the X chromosome.[28] Variants can cause absent or decreased production of dystrophin; an essential protein needed to form and maintain healthy muscle. Individuals with DMD present with symptoms typically around 2 to 5 years of age.[31-33] Initial symptoms include delayed motor milestones, difficulty climbing stairs, waddling, persistent toe walking, elevated creatine phosphokinase, calf hypertrophy, and the classic Gower maneuver.[33-37] As the disease progresses there is symmetric muscle weakness, and the proximal muscles are more affected than the distal muscles. By age 13 years, most individuals utilize a wheelchair full time for mobility needs due to loss of ambulation.[38]

The condition not only affects the skeletal muscle, but also cardiac and respiratory muscle. Involvement of cardiac muscle leads to dilated cardiomyopathy resulting in heart failure. Later stages of DMD might require ventilation support due to breathing difficulty. Cardiopulmonary complications that arise are usually the cause of death within the second to third decade of life.[39] Life expectancy has increased over the years as new treatments emerge, but there is no known cure for DMD.[39-43] Treatments and management guidelines are strictly supportive but can delay disease progression. Treatments such as exon-skipping and stop-codon-read-through therapy are available depending on the individual's genetic variant and stage of disease. Gene therapy is currently being studied at the research level.[44-45]

Grief models developed by Kubler-Ross, Therese Rando, Stroebe and Strut, and Schneider encapsulate the grieving process that parents of children with chronic illness or disability go through. Kubler-Ross's five stages of grief are denial, bargaining, depression, anger, and acceptance.[149] The stage of guilt can sometimes be substituted for bargaining.[156] While this model fits with the end stages of DMD, other grief models have been developed that may be applicable to families during the diagnosis and early stages of DMD. Therese Rando developed a mourning process model consisting of the "Six R's": recognize, react, recollect and/or re-experience, relinquish, readjust, and reinvest.[150] Rando's model can be applied to how parents cope and adjust to their child's diagnosis. Margret Stroebe and Henk Strut's "Dual Process" model of grieving describes how an individual deals with loss by oscillating between the internal loss orientation and restoration orientation. The internal loss state is where the individual focuses on the loss that they are experiencing and the motions that surround it while the restoration state focuses on aspects of life that we need or want.[151] As a result, the loss is processed over time in the capacity at which the individual is capable of while dealing with aspects of everyday life. Lastly, Schneider 1983's model provides a holistic approach to grief that is applicable to families during the diagnosis and early stages of DMD.[152] This holistic approach not only examines how grief affects an individual in the biological, emotional, and behavioral sense, but also one's intellect, spirituality, and attitude. In Schneider's model there six stages of grief: initial awareness, strategies to overcome loss, awareness of loss, completions, resolution, and reformulation, and transcending loss.[152]

Parents of children with Duchenne Muscular dystrophy display a similar experience, if not a more significant level of psychological stress, to parents of children with other chronic illnesses.[190] They also experience five times more stress than parents of children without a chronic illness/disability[191-194,198]. Miller, 1990 demonstrated that the most stressful periods for parents and

families of individuals with Duchene Muscular Dystrophy are the time of diagnosis, loss of ambulation, adolescence, and end stages of the condition.[2] Numerous studies have shown that, overall, parents of individuals with DMD are at an increased risk for developing or experiencing depression or anxiety.[3-4,185-187,195-197] 31-80% of parents report experiencing moderate or severe depression or cried sometimes, often, or always.[3-4,186-187] 21-50% of parents report experiencing moderate to severe anxiety where there is constant worrying.[3,185,187] In addition, 50-60% of parents report poor sleep quality, reduced sleep efficiency, and daytime dysfunction, which can exacerbate mental health symptoms.[185-186,196,201-204]

Studies have shown that parents want support resources to develop better coping skills and parents can cope well, if given the appropriate resources.[183,199,201] Saetrang et al., 2019 found that parents needed professional help to work through the shock of their child's diagnosis.[201] Support resources can encourage coping strategies that parents describe as 'successful' in minimizing distress, such as living in the moment and appreciating present abilities, intellectualization, being proactive, and setting short-term goals.[183,199,201] Mah et al., 2012 states that the need for psychological support may decrease as the disease progresses due to better coping strategies, adjustment, and an accepted reality.[183]

Psychological interventions including psychotherapy, psychiatry, and support groups have shown to be effective in helping those with mental health concerns. According to the American Psychiatric Association psychotherapy is talk therapy with the goal of aiding individuals with mental health conditions or emotional difficulties to minimize symptoms and increase healing and functionality.[205] Psychiatry is a form of medical care in which a medical doctor specializing in mental health examines the mental and physical aspects of mental health conditions to diagnose, treat, and prevent them.[234] Support groups, by definition, are comprised of a group of individuals

who share a similar life stressor, transition, or affliction engaging in mutual support to improve coping and adjustment, alleviate loneliness, facilitate personal empowerment, and offer a sense of community.[271,275,301] Parental experience in support groups has been studied for parents within the Duchenne Muscular Dystrophy community. Kornfeld et al., 1979 found that parents were able to express fears or concerns, learn from each other, and increase their awareness of attitudes toward themselves, their children (with or without DMD), and the world in general.[188] Those who are dissatisfied with offline support are significantly more likely to get support online[269]. The disinhibition effect is the sense of anonymity that puts one at ease reducing the fear of stigmatization.[301] With the positive aspects of support groups there are also negative aspects, such as social comparison,[285] and support groups may not be helpful for all individuals.[290]

Studies conducted in the U.S. as well as across the world show that a majority of individuals experiencing mental health concerns remain untreated or do not seek treatment.[311,313-315] Blumenthal et al., 1996 reports that 45% of individuals seek treatment for their mental health concerns while 56% do not.[315] Since most individuals do not utilize psychological treatments, it is reasonable to question what barriers or reasons prevent individuals from seeking treatment. Typically, the types of barriers can be divided into two categories: attitudinal and structural. Attitudinal barriers are internal factors or reasons that affect utilization of psychological interventions, include negative attitudes towards mental health interventions, believing in handling the mental health concerns alone, low perceived need or severity of concerns, believing treatments would not help, and stigma.[310,315,320-321] Numerous studies have shown that internal beliefs and attitudes affect a person's receptivity to psychological interventions.[315,318-321] Furthermore, mental health symptoms, such as depression, can further exacerbate attitudinal barriers such as lack of motivation, emotional concerns, negative feelings toward therapy, and stigma.[5]

Structural barriers are external reasons that prevent an individual from seeking psychological interventions or having access to psychological interventions. A study conducted on the primary care patient population found that some perceived barriers to psychotherapy include cost, time constraints, transportation difficulties, and childcare or caring for sick/disabled loved ones. The same study also reported that 59.5% of participants stated that at least one of the barriers mentioned would make it difficult to attend and participate in psychotherapy.[5] In the past numerous studies found that financial costs tend to be a major structural barrier due to high costs or lack of insurance coverage.[306-309] Wang et al., 2005 found that those in low-income, low education, urban areas have higher treatment inadequacy.[311] However, the implementation of Health Maintenance Organizations (HMOs) and the Affordable Care Act, individuals have better access to psychological interventions.[302-305] Lastly, for parents of children with a disability or chronic illness, the main structural barrier tends to be the lack of time.[270] Understanding the prevalence of these barriers allows for health care providers as well as public health workers to create ways in which these barriers can be minimized to improve access to psychological interventions especially when there is a clear unmet need for a group of individuals.

This study utilized an online quantitative survey targeted to parents or guardians of individuals diagnosed with Duchenne Muscular Dystrophy to assess the percentage of parents/guardians of individuals diagnosed with Duchenne Muscular Dystrophy who have utilized psychological interventions, such as psychotherapy, psychiatry, or group therapy. In addition, the survey will also examine the barriers or reasons parents/guardians of individuals with Duchenne Muscular Dystrophy chose not to utilize psychological interventions. The results of this survey will help genetic counselors and other healthcare providers to better meet the psychological needs

of parents/guardians of individuals with Duchenne Muscular Dystrophy or other neuromuscular conditions.

3.2 Methods

The University of Pittsburgh Institutional Review Board approved this study (STUDY20100066) as Exempt which met their regulatory requirements. A copy of the IRB approval letter is included in Appendix A.

3.2.1 Study Population

The target population for this research was parents or guardians 18 years of age and older who have a living or deceased child/children with Duchenne Muscular Dystrophy. The families could live anywhere in the world. The only exclusion criterion was if the parent or guardian did not have at least one child with Duchenne Muscular Dystrophy.

3.2.2 Survey Development

The survey was developed in Qualtrics, a web-based service that allows users to easily create a survey, collect and store data securely, analyze responses, and generate graphs of results. This software meets the University Data Security standards. The full survey (Appendix C) consisted of 40 questions, both multiple choice and matrix style. Skip logic was utilized to show participant questions relevant to answers in certain questions answered previously. It contained

seven sections: informed consent, family history, personal psychological history, psychological health screening, utilization of psychological interventions, barriers to psychological interventions, and demographics. The psychological health screening section of the survey was adapted from the Generalized Anxiety Disorder-7 (GAD-7) and Patient Health Questionnaire-9 (PHQ-9) to screen for the presence of clinically significant anxiety disorder or depression, respectively.[326-328] The survey was reviewed and piloted by the thesis committee before distribution and recruitment.

3.2.3 Survey Content

Within the informed consent section (Appendix B), the purpose, goals, risks, and benefits of the study were discussed. There were minimal risks to participants, which could include psychological distress when answering the questions. Participants were informed that their participation was entirely voluntary and anonymous. No identifying information was captured, and participants were able to exit the survey at any point. Email addresses of the principal investigator and faculty mentor were provided should participants have any questions about the study, survey, or results. Lastly, participants were asked to select if they consent to take the survey or not. If they consent to participate, they were able to begin the survey. If they chose not to participate, they were re-directed out of the survey.

The family history section's purpose was utilized to confirm that participants met the inclusion criteria and to trigger the skip-logic feature. The personal psychological history section ascertained whether parents were already utilizing psychological interventions due to a mental health diagnosis. The psychological health screening, utilization of psychological interventions, and barriers to psychological interventions sections had participants think about their experience as a parent of an individual with Duchenne Muscular Dystrophy in two timeframes. Miller, 1990

demonstrated that the most stressful periods for parents and families of individuals with Duchene Muscular Dystrophy are at the time of diagnosis and at times of notable disease progression.[2] Based on this, parents were asked questions regarding their experience during the diagnosis and over the years as the disease progresses. If a family had received a diagnosis within the past 12 months, participants were only shown questions regarding their experience during their child's diagnosis. These screening measures were adapted with the help of a behavioral health specialist on the thesis committee. Participants were then asked if they have ever used certain psychological interventions. If participants answered "no" to utilizing a psychological intervention, they would be asked to select any barriers that prevented them from accessing those interventions. The last section asked participants to answer general demographic information.

3.2.4 Study Recruitment

Recruitment occurred via two methods: in-person and through distribution by advocacy groups. During the recruitment period an IRB approved flyer (Appendix D) was handed out to parents of children with Duchenne Muscular Dystrophy in the Muscular Dystrophy Clinic at Children's Hospital of Pittsburgh. Advocacy groups played a major role during recruitment. An IRB approved email script (Appendix E) along with the IRB approved flyer was emailed to numerous advocacy groups around the world in December 2020 and January 2021. The advocacy groups that agreed to aide in recruitment included Parent Project Duchenne (United States of America and Czech Republic), Therapeutic Research in Neuromuscular Disorders Solutions (TRiNDS), Little Steps Israel, and the Muscular Dystrophy Association (United States of America, Argentina, and Canada). The advocacy groups advertised the survey via social media posts and email list serv. The introductory script also asked participants to forward the survey to other parents

who match the inclusion criteria and who would possibly be willing to complete the survey. The survey remained open from the end of December 2020 to the end of February 2021.

3.2.5 Descriptive Statistics and Thematic Analysis

The results of this study were evaluated using descriptive statistics. Measures of frequency were calculated for each section of the survey. For the demographic, family, and personal history section a total count as well as percent frequency was calculated within the variable such as race, age, and number of children. For the mental health screening participants were placed into categories based on their PHQ-9 and GAD-7 score. The categories were based on guidelines.[326-327] The percent frequency was calculated then displayed within bar graphs. Percent frequency of utilization of psychological interventions was also calculated. The frequency of each barrier selected during the diagnosis stage and disease progression stage was calculated. Then, for each barrier the combined frequency was calculated and displayed in a bar graph. Thematic analysis occurred for participants who wrote in an answer to the selected barrier "Other personal reason." Answers that revealed similar topics, ideas, or language were grouped together.

3.3 Results

A total of 313 participants opened the survey. 33 (10%) participants opened the survey and did not answer any questions and were excluded from analysis. 280 participants opened the survey and answered some or all of the questions. Of those 280, 41 (14.6%) answered some questions but not the required questions, therefore these responses were excluded from analysis. 239 participants

completed the entire survey or all of the required questions. Of the 239 participants, 5 (2%) did not provide consent and 2 (0.8%) declined to participate in this study, therefore these responses were excluded from analysis. Of the 239 participants, 1 (0.4%) completed the required questions, but did not complete the demographic section. Due to this partial data was recorded. To participate in this study, participants were required to have at least 1 child with Duchenne Muscular Dystrophy. Of the 239 participants, 2 (0.8%) did not meet this requirement, therefore they were excluded from analysis. The final total number of participants that were included in the analysis was 230 (73%) individuals.

3.3.1 Demographics

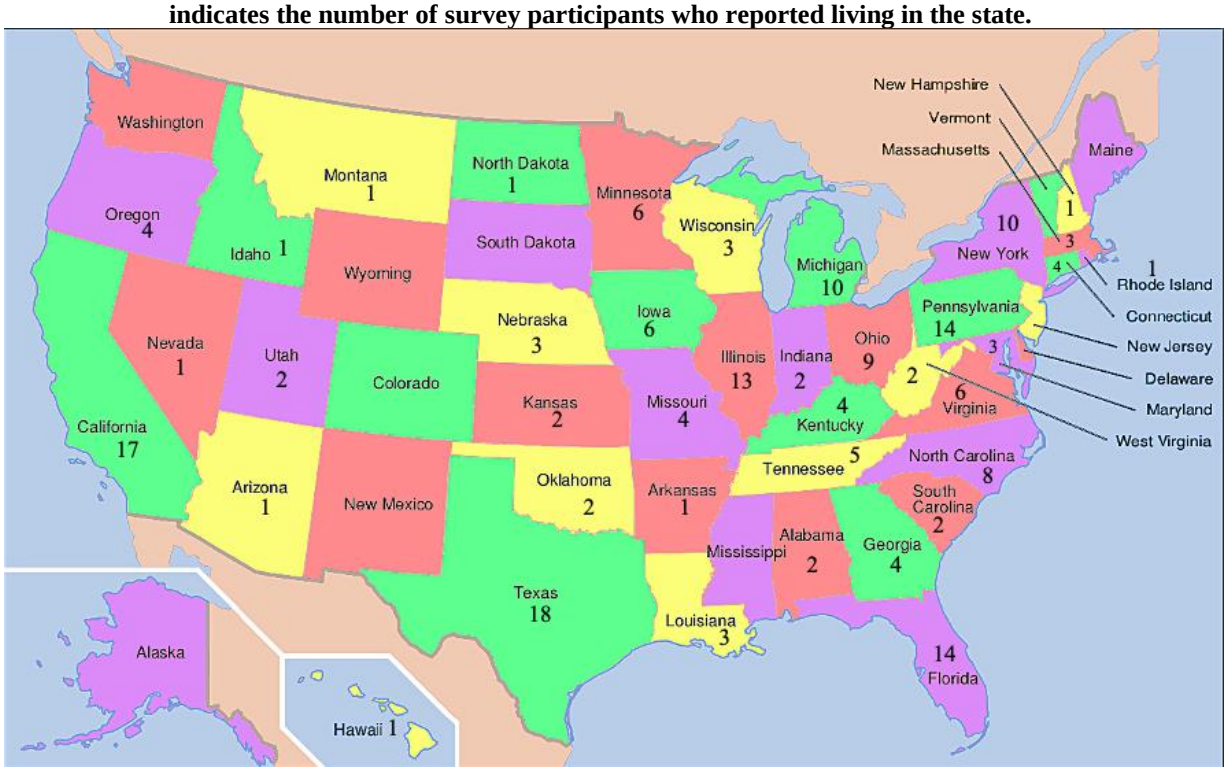
Demographic statistics are detailed in Table 1. The sample population was predominantly female (84.8%), ranged in age from 35-44 years (38.7%), married (72.4%), had some college education (36.4%), has a household income of over $100,000 (37.3%), and identified as White (84.3%). 5 (2.2%) of participants chose “Other/unknown” as their race/ethnicity and 3 of those 5 (60%) people wrote in their answer. Most participants reported “No religious affiliation” (26.1%). 44 participants selected “Other/unknown” under religious affiliation and 39 (89%) participants of those 44 wrote in an answer. Geographically, 96% of participants live in the United States of America followed by Czechia (1.3%), Canada (1.3%), United Kingdom (0.4%), Sweden (0.4%), and India (0.4%). Figure 1 demonstrates the number of participates in each state. The states with the largest number of participants were California, Texas, Florida, and Pennsylvania.

Table 1 Demographic Statistics

Demographic		N (%)
Gender	Female	195 (84.8)
	Male	32 (14.8)
	Non-binary/third gender	0 (0)
	Other	0 (0)
	Prefer not to say	1 (0.43)
Age	18-24 years	0 (0)
	25-34 years	21 (9.1)
	35-44 years	89 (38.7)
	45-54 years	74 (32.2)
	Over 55 years	46 (20)
Race/Ethnicity	White	194 (84.3)
	Black or African American	2 (0.87)
	Asian	9 (3.9)
	Native Hawaiian or Pacific Islander	1 (0.4)
	Hispanic or Latino	23 (10)
	American Indian or Alaska Native	1 (0.4)
	Native American	0 (0)
	Two or more	4 (1.7)
	Other/Unknown	5 (2.2)
Religious Affiliation	No religious affiliation	60 (26.1)
	Protestant	43 (18.7)
	Catholic	54 (23.5)
	Mormon	2 (0.8)
	Jewish	2 (0.8)
	Hindu	5 (2.2)
	Buddhist	1 (0.4)
	Muslim	3 (1.3)
	Other/unknown	44 (19.1)
	Prefer not to say	14 (6.1)
Marital Status	Single- never married	14 (6.14)
	Married	165 (72.4)
	In a domestic partnership	6 (2.6)
	Divorced	31 (13.6)
	Divorced and remarried	8 (3.5)
	Widowed	4 (1.8)
Level of Education	High School or less	25 (11)
	Some College	83 (36.4)
	Bachelor's degree	64 (28.1)
	Master's degree or higher	56 (24.6)
Household Income	Less than $20,000	18 (8.3)
	$20,000-$34,999	23 (10.6)
	$35,000-$49,999	20 (9.2)
	$50,000-$74,999	33 (15.2)

	$75,000-$99,999	42 (19.4)
	Over $100,000	81 (37.3)
Country	United States of America	216 (96)
	United Kingdom	1 (0.4)
	Sweden	1 (0.4)
	India	1 (0.4)
	Czechia	3 (1.3)
	Canada	3 (1.3)

Figure 1: Participant Demographics in the United States of America. A number by the state name indicates the number of survey participants who reported living in the state.



3.3.2 Family and Personal History

230 participants were asked questions regarding their family history and personal psychiatric history (Table 2). A majority of the sample population had a total number of 2-4 children (77%) followed by 1 child (17.9%) and more than 4 children (5.2%). One of the inclusion criteria for the study was that the participants must have at least one child with Duchenne Muscular Dystrophy (DMD). This study's sample population had either 1 (88%) or 2 (12%) children with

DMD. Before having a child diagnosed with DMD, a majority of parents did not have any children (48.3%). Parents were typically between the ages of 24-35 years old (57%) when their child with DMD was born and 93.5% of participants reported that their child had not been diagnosed with DMD within the past 12 months.

Participants were then asked questions regarding their personal psychiatric history. 80% of participants reportedly did not have a mental health diagnosis before they had their child with DMD. If participants answered "Yes" to having a mental health condition before their child was diagnosed with DMD, they were asked to list the name of their mental health condition. Twenty participants listed "Anxiety and Depression", twelve listed "Depression", five listed "Anxiety", and six participants had other mental health conditions. Most participants also reported that they had not utilized psychotherapy (mental health counseling) (72.2%) or psychiatry (86%) before their child with DMD was born.

Table 2 Family and Personal History Statistics

Category		**N (%)**
Total number of Children	1	41 (17.9)
	2-4	177 (77)
	More than 4	12 (5.2)
Number of Children Diagnosed with DMD	1	202 (88)
	2	28 (12)
	3 or more	0 (0)
Number of Children before having a child with DMD	None	111 (48.3)
	1	69 (30)
	2	34 (14.8)
	3 or more	16 (7)
Child diagnosed within the past 12 months	Yes	15 (6.5)
	No	215 (93.5)
Age of parent when oldest child with DMD was born	Under 18 years	3 (1.3)
	18-24 years	31 (13.5)
	25-34 years	131 (57)
	35-44 years	58 (25.2)
	45-54 years	7 (3)
	Over 55 years	0 (0)
Mental Health diagnosis before child was diagnosed with DMD	Yes	45 (20)
	No	185 (80)

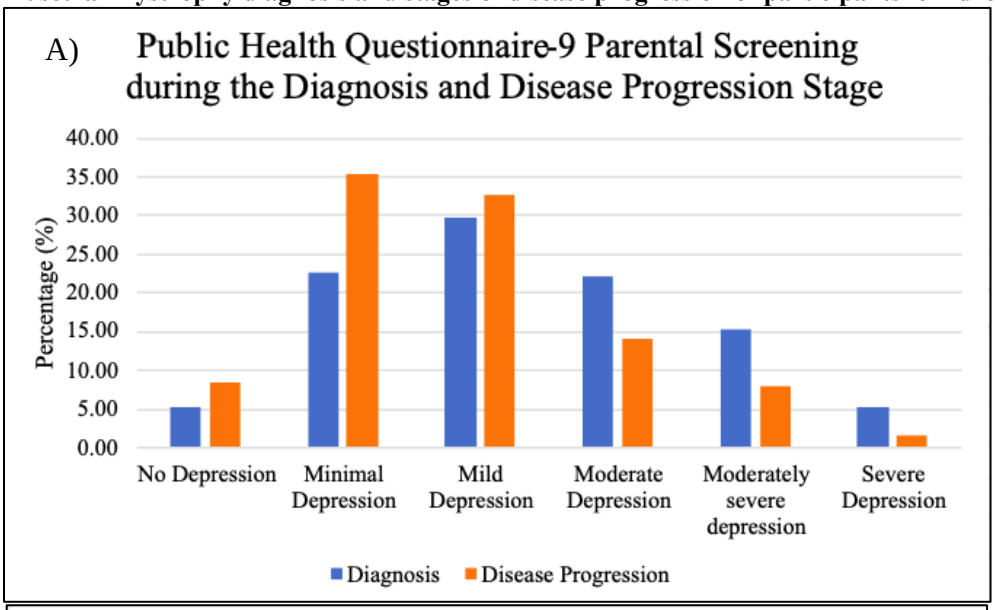
Mental Health Counseling utilization before child with DMD	Yes	64 (27.8)
	No	166 (72.2)
Psychiatry utilization before child with DMD	Yes	32 (14)
	No	198 (86)

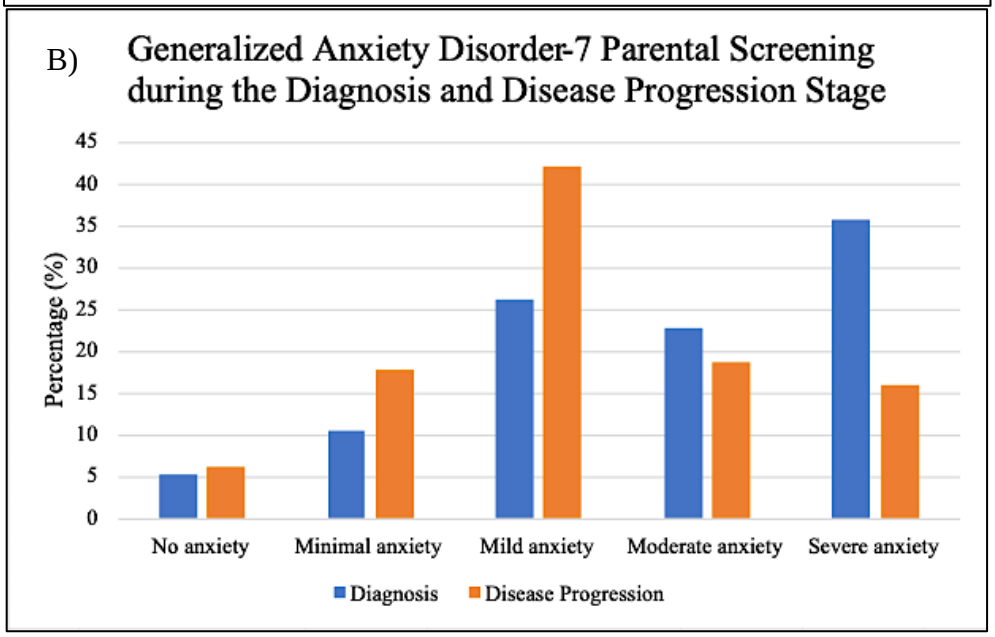
DMD: Duchenne Muscular Dystrophy

3.3.3 Mental Health Screening

230 participants participated in mental health screening via the Public Health Questionnaire-9 (PHQ-9) and the Generalized Anxiety Disorder-7 (GAD-7). All participants answered screening questions for the time frame of their child's diagnosis. 215 participants answered screening questions for the time frame of their child's disease progression because they reported that their child was not diagnosed with DMD within the past 12 months. Figure 2A shows the results of the PHQ-9 screening during both the diagnosis and disease progression stage. During the diagnosis stage 94.78% of participants experienced varying degrees of depression symptoms with 42.6% experiencing moderate to severe depression. 5.22% had reported no symptoms of depression. During the disease progression stage, lower rates on depression occurred with 91.63% experiencing some level of depression and 23.26% experiencing moderate to severe depression. Figure 2B shows the results of the GAD-7 screening. 93.95-94.78% of participants experienced anxiety symptoms of varying severity. Participants experienced higher rates of moderate to severe anxiety during the diagnosis stage (58.26%) compared to the disease progression stage (34.41%). Overall, anxiety symptoms, especially moderate to severe, were more prevalent compared to depression symptoms.

Figure 2: Participant Mental Health Screening: A) Public health questionnaire-9 mental health screening for depression during the Duchenne Muscular Dystrophy diagnosis and stages of disease progression of participants' children. B) Generalized anxiety disorder-7 mental health screening for Anxiety during the Duchenne Muscular Dystrophy diagnosis and stages of disease progression of participants' children.





3.3.4 Psychological Intervention Utilization

Participants were asked about their utilization of psychological interventions during the diagnosis stage of their child's condition as well as when the child's condition has progressed over the years (Table 3). All 230 individuals were asked if they utilized psychological interventions during the diagnosis stage of their child's condition. 215 participants were additionally asked about their utilization of psychological interventions as their child's condition has progressed over the years. During the diagnosis stage the majority of participants did not utilize psychotherapy (67.4%), psychiatry (85.7%), in-person support groups (77.8%), or online support groups (78.3%). The same occurred for utilization of psychological interventions as the child's condition progresses. The majority of parents did not utilize psychotherapy (56.7%), psychiatry (80%), in-person support group (72.6%), or online support groups (67%). The proportion of individuals who did utilize each intervention was greater during the disease progression stage (20%-43.3%) compared to the diagnosis stage (14.3%-32.6%).

Table 3 Psychological Intervention Utilization

Intervention		**Diagnosis**	**Disease Progression**
		N (%)	**N (%)**
Psychotherapy (Mental Health Counseling)	Yes	75 (32.6)	93 (43.3)
	No	155 (67.4)	122 (56.7)
Psychiatry	Yes	33 (14.3)	43 (20)
	No	197 (85.7)	172 (80)
In-Person Support Group	Yes	51 (22.2)	59 (27.4)
	No	179 (77.8)	156 (72.6)
Online Support Group	Yes	50 (21.7)	71 (33)
	No	180 (78.3)	144 (67)

3.3.5 Barriers to Psychological Interventions

When participants answered "No" to any of the psychological intervention utilization questions, they were asked what barriers prevented them from accessing that psychological intervention. A list of barriers was provided, and participants were asked to check all that applied to them. Table 3 and Figure 3 describe the frequency at which each barrier was chosen. The top 3 barriers selected for psychotherapy were that participants did not feel the need to utilize the intervention (n=142), financial reasons (n=78), and time constraints (n=77). Similarly, the top 3 barriers to psychiatry were that participants did not feel the need to utilize the intervention, time constraints (n=83), and financial reasons (n=80). For both psychotherapy and psychiatry "I felt that I did not need to" was the most common reason selected. Participants not receiving a referral to the intervention was the fourth most frequent barrier for both psychotherapy (n=60) and psychiatry (n=70).

The 3 most common barriers for in-person and online support groups were a lack of local support group available (n=150), participants felt that they did not need a support group (n=131), and time constraints (n=85). Similar results occurred for online support groups where a lack of local support group available (n=131), participants felt that they did not need a support group (n=133), and time constraints (n=61) were the barriers most frequently chosen. The frequency at which a single participant chose multiple barriers was 105, 103, 84, and 72 for psychotherapy, psychiatry, in-person support groups, and online support groups, respectively. Overall, across all psychological interventions the predominating barriers or reasons for not utilizing an intervention were "I felt that I did not need to" and the lack of support groups available.

In addition, participants had the option to write in a barrier or reason for not utilizing a psychological intervention if they chose the option "Other personal reason". Table 5 shows the

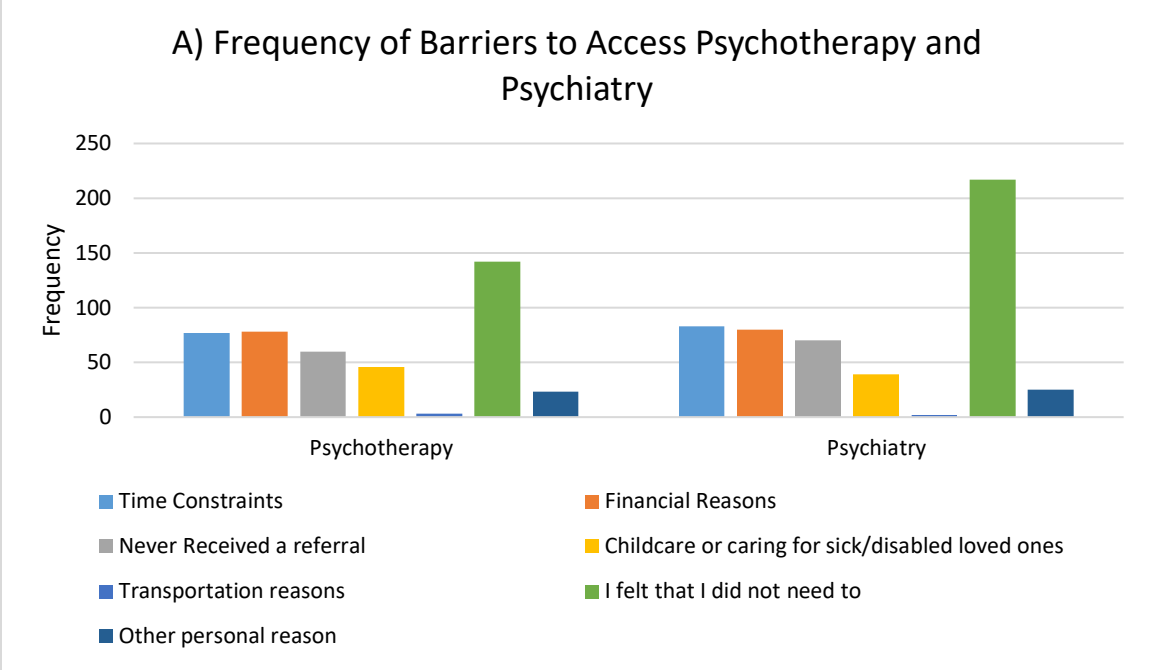
qualitative themes that arose from participants' answers. Common themes for barriers shared across all four types of psychological interventions include: being emotionally overwhelmed, having other sources of support, the COVID-19 pandemic, and lack of resource information or availability. Psychiatry revealed to have alternative access with participants being prescribed medications by their primary care physician rather than a psychiatrist. Both psychotherapy and psychiatry had barriers due to stigma. Lastly, both in-person and online support groups had barriers due to previous experiences being either not helpful or poor.

Table 4 Barriers to Psychological Interventions

Intervention	Barrier	Diagnosis	Disease Progression	Total
		N	N	N
Psychotherapy (Mental Health Counseling)	Time Constraints	45	32	77
	Financial Reasons	42	36	78
	Never received a referral	35	25	60
	Childcare or caring for sick/disabled loved ones	26	20	46
	Transportation reasons	2	1	3
	I felt that I did not need to	84	58	142
	Other personal reason	13	10	23
	Multiple barriers chosen	60	45	105
Psychiatry	Time Constraints	43	40	83
	Financial Reasons	41	39	80
	Never received a referral	37	33	70
	Childcare or caring for sick/disabled loved ones	20	19	39
	Transportation reasons	1	1	2
	I felt that I did not need to	122	95	217

	Other personal reason	14	11	25
	Multiple barriers chosen	52	61	103
In-person Support Group	Time Constraints	43	42	85
	Financial Reasons	21	22	43
	Lack of local support group available	79	71	150
	Childcare or caring for sick/disabled loved ones	22	26	48
	Transportation reasons	4	3	7
	I felt that I did not need to	69	62	131
	Other personal reason	24	15	39
	Multiple barriers chosen	33	51	84
Online Support Group	Time Constraints	32	29	61
	Financial Reasons	16	15	31
	Lack of support group available	73	58	131
	Childcare or caring for sick/disabled loved ones	15	12	27
	Transportation reasons	2	2	4
	I felt that I did not need to	68	65	133
	Other personal reason	24	20	44
	Multiple barriers chosen	34	38	72

Figure 3: Frequency of barriers selected for not utilizing psychological resources. A) The frequency at which each barrier to accessing psychotherapy and psychiatry was chosen. B) The frequency at which each barrier to accessing in-person and online support groups was chosen



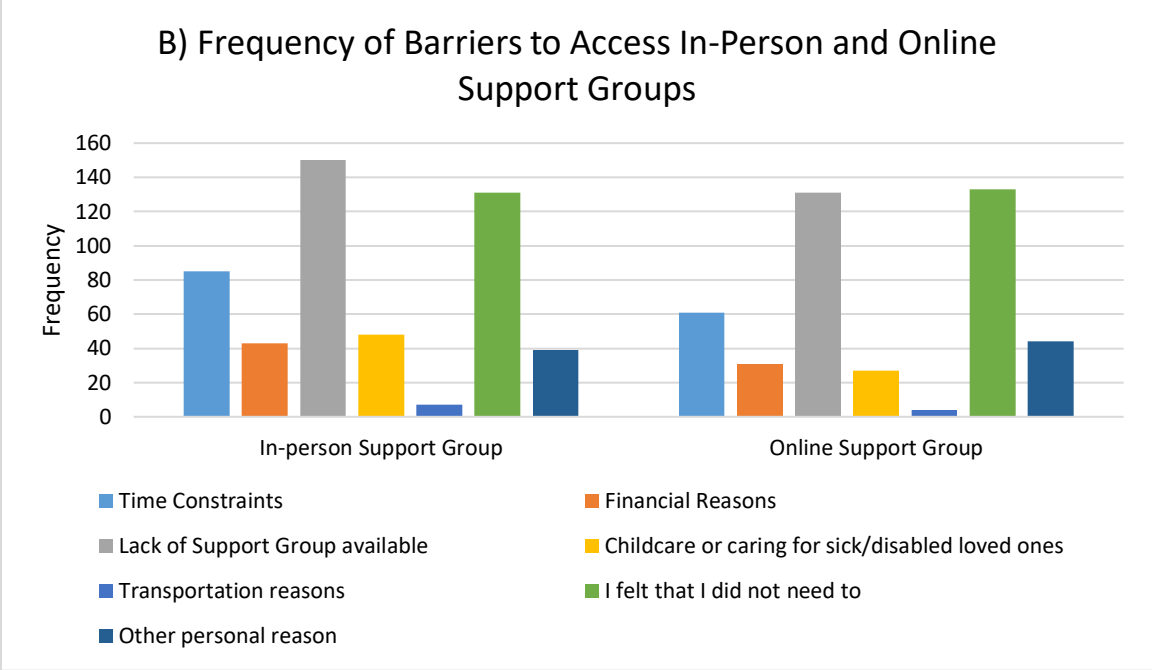


Table 5 Thematic Analysis of "Other personal reason" responses

Intervention	Theme	Quotation
Psychotherapy (Mental Health Counseling)	Other Support Available	1. Seen my preacher 2. I had family to talk to.
	Emotionally Overwhelmed	1. Was still trying to process everything and wasn't emotionally prepared to be attentive to my needs. 2. Overwhelmed with diagnosis and formulating a plan
	Lack of resource availability	1. COVID-19 2. Stationed overseas, was not an option 3. Lack of specialists. Long waiting lists.
	Personal focus was not on self-care	1. It never crossed my mind; I was focused on getting a care team set up for my son. 2. I had to choose between therapy for myself, or supportive therapies for my son. I chose my son's therapies. 3. I felt it wouldn't help since curing DMD was the answer 4. Younger siblings to care for.
	Stigma	1. Negative impact on employment
Psychiatry	Other Support Available	1. Had a strong faith support system 2. I would pray a lot and God would help me. 3. Seen my preacher 4. I saw a counselor and that helped.
	Alternate access	1. Antidepressants are available via primary MD not psych. 2. I was prescribed medication by my primary care 3. Went to see PCP instead 4. Received prescription from primary care physician
	Lack of resource availability	1. COVID-19 2. Psychiatrists are so hard to see. They're always booked so long out. 3. Lack of specialists. Long waiting lists. 4. It's too hard to get booked in a timely manner with one.
	Past bad experience	1. I only saw a psychiatrist in my teen years, and she misdiagnosed me and gave me drugs I was allergic to which I stopped after a few days. I have never seen a psychiatrist since.
	Personal focus not on self-care	1. It never crossed my mind; I was focused on getting a care team set up for my son.
	Stigma	1. Negative impact on employment

		2. I did not need addicted to antianxiety medicine.
In-person Support Group	Pandemic (COVID-19)	1. Pandemic limited in-person meetings. 2. Coronavirus 3. Covid-19 4. Covid and time constraints
	Other Support Available	1. Seen my preacher 2. I attend support groups, none related to DMD
	Lack of resource availability	1. There are no group therapy's available specifically for Duchenne 2. Difficult to access 3. I attend support groups. None related to DMD because there are none.
	Lack of resource information	1. Was unaware of resources 2. Didn't know how to access relevant group 3. Didn't know where to look
	Overwhelmed emotionally	1. I wasn't ready to hear other parents' stories. I needed to come to grips with the diagnosis on my own first as I had just recently become engaged, and my children were dealing with issues regarding their father's (former husband) behavior toward them. 2. I wasn't ready to hear more about the disease at the time of diagnosis. 3. Denial 4. I feel it would raise my anxiety level. 5. Without a diagnosis I did not think a support group existed. Just after diagnosis we were too busy coping to look for a group, but we did get a call from a PPMD rep, so we talked one on one by phone and then our family attended a PPMD conference about a year after dx
	Personal focus not on self-care	1. Not enough time in my schedule to dedicate to an in-person support group. I need flexibility since I am caring for my son. 2. Other children I needed to care for. 3. DMD parents need better treatments and or a cure for Duchenne
	Past Experience was not helpful	1. Did not feel like it helped. Other people in the group were in worse mental shape then I was. 2. the one we did attend was not helpful, too depressing 3. I also don't feel most people can relate. And I don't fit into most of the DMD mom groups.
	Miscellaneous	1. Not interested
Online Support Group	Other Support Available	1. Seen my preacher 2. Only chat boards available

	Lack of resource availability	1. There was no group for us parents to attend. I asked. 2. Wasn't available
	Lack of resource information	1. Online? I was unaware of the internet and social media was barely invented! 2. Didn't know where to look 3. was unaware of resources 4. I have not found or searched for online DMD support group.
	Overwhelmed emotionally	1. I wasn't ready 2. I wasn't sure what to do during that time. I felt numb during my son's diagnosis. 3. Denial 4. I wasn't ready to hear more about the disease at the time of diagnosis. 5. I feel it would raise my anxiety level 6. I wasn't ready to hear other parents' stories. I needed to come to grips with the diagnosis on my own first as I had just recently become engaged, and my children were dealing with issues regarding their father's (former husband) behavior toward them. 7. I'm afraid it would be too overwhelming for me.
	Past Experience was not helpful	1. Too difficult seeing my future with the disease. Most parents had children further along than my son 2. I feel I don't fit in, although the chat on FB has been helpful on criticism with the terrible help we get from MDA. I don't feel crazy/alone on this 3. DMD online support groups are helpful with feelings of not being alone BUT continued & severe feelings of grief for other people's DMD son's compounds things needlessly 4. don't like online support 5. I also don't feel most people can relate. And I don't fit into most of the DMD mom groups.
	Miscellaneous	1. not really interested 2. Not interested in online group 3. Child's doctor recommended not utilizing online resources. 4. Support groups too far away to attend

DMD: Duchenne Muscular Dystrophy

3.4 Discussion

3.4.1 Mental Health Screening

This is not the first study examining rates of depression or anxiety in parents of individuals with Duchenne Muscular Dystrophy. For our study it was not one of the main goals to capture the level of depression or anxiety symptoms in parents of individuals with DMD, but to use the information to provide context to the patterns discovered for the utilization of and barriers to psychological interventions. Miller, 1990 demonstrated that the most stressful periods for parents and families of individuals with Duchene Muscular Dystrophy are the time of diagnosis, loss of ambulation, adolescence, and end stages of the condition, therefore we asked participants to think about those time periods when they answered mental health screening questions.[2] Figure 2A shows the percentage of participants that experienced varying degrees of depression symptoms during the diagnosis and disease progression stage. During the diagnosis stage 94.78% of participants experienced some degree of depression symptoms. During the disease progression stage 91.63% of participants experienced some degree of depression symptoms. Previous studies have reported that 50-80% of parents of individuals with DMD experience depression symptoms.[3,186-187,196] Magliano 2014 found that parents of individuals with DMD reported that they felt depressed or cried sometimes, often, or always.[186] In addition, Landfeldt 2016 found that 50% of parents of individuals with DMD were moderately or extremely depressed.[187] Our results trended higher when comparing our results for overall depression symptoms to the results of those studies. However, our results were lower compared to previous studies for moderate to severe depression (42.6% during diagnosis and 23.26% for disease progression). This leads to the conclusion that

parents do experience some form of depression, most often in the minimal to mild range, but not on the severe end.

In terms of anxiety, Figure 2B revealed that 94.78% and 93.95% experienced varying degrees of anxiety during the diagnosis and disease progression stage, respectively. Previous studies have reported that parents of individuals with DMD experience 21-50% of anxiety symptoms[3,185,187]. Again, Landfeldt 2016 reported that 50% of parents of individuals with DMD were moderately or extremely anxious.[187] Our study found that 58.26% and 34.41% experienced moderate to severe anxiety during the diagnosis stage and disease progression stage respectively. Our results reported higher anxiety prevalence overall and higher moderate to severe anxiety, but only during the diagnosis stage. Based on these results it can be concluded that the participating parents experienced a high level of anxiety, especially more severe anxiety during the diagnosis stage. Overall, there was a higher prevalence of depression and anxiety symptoms found during the diagnosis stage compared to the disease progression stage. An explanation for this phenomenon could be that within the time after the diagnosis, parents are allowed space to grieve and cope thus reducing the intensity of mental health symptoms. This would result in the lower scores on the PHQ-9 and GAD-7.

3.4.2 Utilization of Psychological Interventions

To our knowledge this is the first study to examine the utilization rates of psychological interventions specifically for parents in the Duchenne Muscular Dystrophy community. Due to this our study will compare psychological interventions, such as psychotherapy, psychiatry, and both in-person or online support groups, to broader target populations across the world. As seen in Table 3, for each psychological intervention there was underutilization during both the diagnosis

stage and disease progression based on the levels of depression and anxiety reported by the respondents. This is consistent with many studies worldwide that have reported underutilization of mental health services and high levels of mental health concerns that remain untreated.[311,313,347-348] The World Health Organization (WHO) world mental health survey found that 35.5-50.3% of mental health cases in developed countries and 76.3-85.4% of mental health cases in less-developed countries are not receiving treatment.[313] Wang et al., 2005 reported that 41.1% of individuals in the United States received some type of psychological treatment.[311] Our study's findings fall within the limits or below the results of previous studies. During the diagnosis stage the majority of participants did not utilize psychotherapy (67.4%), psychiatry (85.7%), in-person support groups (77.8%), or online support groups (78.3%). The same occurred for utilization of psychological interventions as the child's condition progresses where parents did not utilize psychotherapy (56.7%), psychiatry (80%), in-person support group (72.6%), or online support groups (67%) (Table 3). This supports the conclusion that parents of individuals with Duchenne Muscular Dystrophy underutilize psychological interventions as a whole.

Taking a deeper look at utilization rates, the proportion of individuals who did utilize each intervention was greater during the disease progression stage (20%-43.3%) compared to the diagnosis stage (14.3%-32.6%). This was surprising because there was a higher prevalence of depression and anxiety symptoms found during the diagnosis stage compared to the disease progression stage within the mental health screening section of this study. Kerr et al. 2000 showed that the need for support typically lessens a few years after the diagnosis stage.[169] However, it is also recognized that the stages of grief are not linear and there is always a possibility to trigger any stage within the grief model as new life challenges emerge.[149-152] The concept of chronic sorrow could also apply, thus introducing a chronic need for psychological interventions to be

implemented at any point.[162] This means that there is always a possibility of parents needing some type of psychological intervention due to the life-limiting nature of their child's condition. Healthcare providers should be aware of these patterns so that psychological interventions are offered or discussed not only during the diagnosis stage of the parents' child's DMD, but also as the condition progresses.

In addition, our study found that the intervention with the greatest proportion of participants utilizing it was psychotherapy (32.6%-43.3%) followed by in-person support groups (22.2%-27.4%), online support groups (21.7%-33%), and psychiatry (14.3%-20%). Numerous studies have shown that individuals experiencing mental health symptoms prefer psychotherapy over psychiatry and if they preferred psychotherapy, they preferred group psychotherapy over individual.[206,308,316,322-325] Kovess-Masfety et al. 2007 looked at mental health preferences in six European Countries and found that psychotherapy was preferred over psychiatry in Belgium, Germany, and the Netherland while France, Italy, and Spain have higher utilization rates for psychiatry over psychology.[350] Mack et al., 2014 discovered that the utilization pattern of psychological services in Germany was highest for psychotherapy, followed by psychiatry, and then self-help groups.[347] Other studies found that 18.8%, 17%, and 40-66.4% of individuals had utilized psychiatry, psychotherapy, and attended a support group respectively.[272,348-349] Our results are consistent with the conclusion that utilization frequency of psychotherapy is greater than psychiatry, but it is surprising that support group utilization was not higher. Reasons for this finding will be discussed further in the next section where we discuss the barriers/reasons participants in this study did not utilize psychological interventions.

3.4.3 Barriers to Psychological Interventions

This study identified a wide variety of barriers and/or reasons that may help to explain the lack of utilizing psychological interventions. There was a mix of attitudinal barriers and structural barriers. For all the psychological interventions the most prevalent reason chosen was the attitudinal barriers "I felt that I did not need to". There could be a few explanations for this phenomenon. The first is that participants truly did not need to use the psychological intervention, however the results of the mental health screening contradict this explanation. The fact that a majority of participants experienced high levels of anxiety suggests that parents could benefit from psychological intervention.

In light of this the other explanation could be the concept of low perceived need along with underlying attitudinal barriers. Numerous studies have found that low perceived need and attitudinal barriers are significant factors in help-seeking behaviors especially for mild to moderate mental health symptoms.[310,334-337] Many community-based surveys have also shown that a majority of individuals worldwide are unable to recognize mental health conditions.[338-343] In addition, it is important to note that due to this phenomenon, individuals can often deem depression symptoms as life stressors which results in a lack of help-seeking behavior. Jorm, Kelly et al., 2006 found that when individuals mislabel depression symptoms this way that they were more likely to believe that the issue could be dealt with without psychological interventions.[344] The combination of low perceived need and low mental health literacy makes a compelling case for why this choice was most prevalent. It is also important to keep in mind that participants could choose multiple barriers, so there were instances where other barriers were chosen along with "I felt that I did not need to". This indicates that the reason for not utilizing psychological interventions is more complex than just a single barrier.

The second major barrier was the fact that there was a lack of support groups available. This finding is more structural in nature and represents a significant unmet need for parents of individuals with DMD. While creating a local in-person support group sounds like a simple solution, the process of developing a support group takes a time commitment not only from physicians and clinicians involved, but the families as well. It would also require resources that neuromuscular clinics might not have. Advocacy groups such as the Muscular Dystrophy Association does have community events, but not a formal support group available. Advocacy groups, neuromuscular clinics, and parents in the community would need to work together if an in-person support group in local areas is desired. Online support groups might be a better solution. There are probably numerous online support groups, but individuals may not know where to look or what organizations are credible. This is where clinicians involved in the care of an individual with DMD should guide parents to credible advocacy groups. In addition, since the use of teleconferencing has increased due to the pandemic (COVID-19), advocacy groups could find creative ways to use the technology for face-to-face support.

The third most common barrier was time constraints. This finding was not a surprise given the fact that previous studies have reported that parents have reported a lack of time to perform daily activities or hobbies.[185-187,270] With the lack of time to be physically present at the psychological intervention, parents in this community might find that telemedicine or a support group that meets virtually would be helpful. Financial barriers do not seem to be a major concern with this cohort, but this is not surprising given that a majority of our cohort has a household income of over $100,000 (Table 1), however financial barriers still made the top three barriers for psychotherapy and psychiatry. Previous studies have shown financial barriers to be among the top structural barriers for psychotherapy and psychiatry and being in a low socioeconomic area can

exacerbate this barrier to an even greater extent.[5,309,311] Financial barriers could also occur due to varying insurance coverage. Having high insurance coverage from the Affordable Care Act or being a part of a health maintenance organization (HMO) leads to better access to mental health services, in this case psychotherapy and psychiatry.[302-307] Support groups are generally free of charge; therefore, it is reasonable for the frequency of financial barriers for support groups to be low.

The fourth major barrier for psychotherapy and psychiatry was that participants had never received a referral. It is known that parents of children with DMD are more likely to experience psychological distress than parents who do not have a child with DMD, therefore they should be linked to appropriate psychological interventions[4]. Mandell et al., 2007 found that parents were more likely to attend support groups if the clinician who diagnosed their child referred them to one, highlighting the importance of counseling by the clinician on support options.[349] Previous studies have shown that physicians fail to recognize symptoms in order to make a referral for treatment for 30 to 50% of individuals with mental health concerns.[329-332] Even when physicians refer individuals for mental health treatment, only 20% of them follow through utilizing the intervention.[316-317] These numbers could be even lower for parents in the DMD community since their child is typically the focus of the doctor's appointment. In the study conducted by Saetrang et al. 2019, one parent described how the sorrow comes on suddenly causing immense exhaustion and feeling alone in grief.[201] This parent recognized that both the child and the parent themselves should receive referrals for mental health treatment to deal with the life-limiting aspect of their child's condition as it progresses, but they did not feel that they were in a position to bring up the concerns during their child's medical appointment. Therefore, even if there is a structural barrier there is a chance of an attitudinal barrier occurring that could affect utilization rates.

Lastly, Table 5 shows similar qualitative themes under the quantitative selection of "Other personal reason". Having other support available or alternative access was a common theme across all psychological interventions. Some studies have shown that parents of individuals with chronic illness or disabilities can adapt well due to family support, religious support, primary care treatment of depression, and other resources.[345-346] Yamaguchi et al., 2019 found that parents want a support person but preferred family, friend, or spouse over a psychological counselor.[333] The qualitative results found similar reasons to these studies. These reasons were mentioned within the selection of "Other personal reason" which was the least frequently selected barrier. Therefore, these findings represent the small portion of individuals who do well adapting without psychological interventions. Other common themes were the effects of the pandemic (COVID-19), parents focus not on self-care, past experiences not being helpful, and being emotionally overwhelmed. It is important to note that the pandemic during the year 2020 to the present has affected the availability of mental health services and ability to access those services due to social distancing and the presence of lockdowns. Therefore, it was not surprising that the effects of the pandemic appeared in the survey results. The themes of being emotionally overwhelmed and the personal focus not being on self-care give further evidence for the underutilization of psychological interventions. It shows that although a majority of parents experience psychological distress it is possible that some parents perceive using resources such as supports groups could increase their distress. Overall, the qualitative results provide details that highlight barriers not available to be selected for a small portion of participants.

3.4.4 Generalizability of Results, Limitations, and Future Research

With the demographics of this study's cohort, there is some generalizability under the assumption that the participants share a similar, if not identical, identification of their race/ethnicity with their child who has DMD. A majority of this cohort identifies as White (84.3%) followed by Hispanic or Latino (10%), Asian (3.9%), Other/Unknown (2.2%), Two or more races/ethnicities (1.7%), Black or African American (0.87%), Native Hawaiian or Pacific Islander (0.4%), and American Indian or Alaska Native (0.4%) (Table 1). A previous cross-sectional study examining prevalence of DMD in the United States of America between 1991–1995, 1996–2000, 2001–2005, and 2006–2010 across various races/ethnicities revealed that prevalence of DMD was highest amongst individuals that identified as Hispanic compared to individuals that identified as non-Hispanic white or black.[25] However, a recent study retrospectively looking at cases between 2006-2015 showed that Duchenne Muscular Dystrophy is significantly more prevalent among individuals who identify as non-Hispanic whites compared to other races and ethnicities.[358] Our study's participants mainly identified as White, similar to Salzberg's study, but the prevalence of DMD amongst minority populations was lower compared to Salzberg's study.[358] This eliminates generalizability due to the lack of racial/ethnic diversity. On the other hand, this cohort is mostly from the United States (96%) and the prevalence data is from research conducted in the United States of America, therefore the results and conclusions from this study have the potential to be generalized to the Duchenne Muscular Dystrophy community within the United States of America but not across race/ethnicity.

It is important to understand the results of this study in the context of its limitations. The first limitation is that for the mental health screening the prompt is asking about symptoms during a time that might have occurred in the past since a majority of parents (93.5%) did not have a child

who was diagnosed with DMD within the past 12 months. The intensity of mental health symptoms during a stressful period could be different from the reflection of that time period. In addition, those diagnosed with a mental health condition (20%) could be more likely to screen positive for depression or anxiety if their condition is currently not well managed and vice versa if the condition is well managed. The second limitation is that the survey was only available in the English language and was not translated even though recruitment emails were sent to DMD affiliated organizations worldwide. Some advocacy groups such as Parent Project Duchenne in Argentina did aide in distributing the survey to members with English literacy, but not having the survey in their language could have affected a participant's ability to complete or fully comprehend the survey. The third limitation is that this study's cohort was gathered through advocacy groups, indicating that they had some level of support and involvement in the Duchenne Muscular Dystrophy community and were more likely to participate in this study. This can cause voluntary response bias where those who choose to participate are different than those who choose not to participate. This can cause an underrepresentation of individuals who might not feel as strongly regarding the study's subject within the DMD community. Overall, there might be unmet need or identification of barriers within the DMD community that this sample cohort was not able to capture. Our data is one set of information. It is entirely possible that with a larger cohort the results might fluctuate.

Regarding future research there are multiple opportunities to expand on the results from this study. Since the majority of participants were from the United States of America, this study can be repeated in other locations across the world. Having the survey translated and increased recruitment and survey completion strategies may allow more demographic diversity. With the survey being translated into other languages, other countries could participate. The main method

for recruitment was through advocacy groups and the survey was to be completed online. Using different recruitment and survey completion strategies would help capture individuals who are not involved with advocacy groups or do not have access to computers.

The survey can also be used as a baseline for expanding this study. Future studies could be done to gather qualitative data to understand the underlying reason for "I feel that I did not need to" response being in the top three reasons for not utilizing psychological interventions. It would be interesting to examine if the qualitative study would produce similar results to the small portion of qualitative data gathered in this study.

Lastly, based on these results future research should examine how public health interventions, such as implementing mental health screenings in the neuromuscular clinic for both parents and children and mental health literacy campaigns, effect utilization rates of psychological interventions. Wells et al. 2000 found that training healthcare workers to routinely screen and discuss mental health treatment improved utilization rates as well as mental health outcomes for patients.[357] Individuals who are informed about mental health resources are more inclined to use them.[206] These methods could help reduce attitudinal barriers. Ways to improve structural barriers would be to implement a support group or encourage use of telemedicine options for psychotherapy and psychiatry. Numerous studies have shown the effectiveness of telemedicine for mental health.[352-356] Even psychotherapy over the telephone could reduce barriers for those who do not have access to the internet. Brenes et al. 2015 found the telephone-delivered cognitive behavior therapy was able to significantly decrease generalized anxiety symptoms, depressive symptoms, and worry severity.[351] Overall, this study has the capability to be the foundation for future research regarding the psychosocial health of families within the Duchenne Muscular Dystrophy community.

3.5 Conclusion

Overall, there is an underutilization of psychological intervention for parents in the Duchenne Muscular Dystrophy community. The fact that there is underutilization is interesting given the fact that a majority of participants experience varying degrees of depression and anxiety during their child's diagnosis stage and disease progression stage. The percentage of individuals experiencing moderate to severe anxiety was higher than the percentage of individuals experiencing moderate to severe depression. The mental health screening confirms what previous studies have found and counseling on appropriate mental health therapies should occur during the child's neuromuscular clinic appointment.

It was discovered that the underutilization of mental health services was explained by a variety of barriers. Attitudinal barriers dominated over structural barriers for psychotherapy and psychiatry. The high frequency of the attitudinal barrier could be explained by low perceived need or low mental health literacy in the context of the mental health screening results. Physicians involved in the child's multi-disciplinary team should utilize family-centered care and implement mental health screening tools like the ones used in this survey.

For support groups the biggest barrier is the lack of support group availability. This is a need that should be recognized among DMD related organizations now that these results are available. With the effects of the 2020 pandemic, virtual video communication has increased. With new platforms being available because of the pandemic, the DMD organizations have a unique opportunity to attend to this unmet need. Unsurprisingly time constraints, financial constraints, and lack of referrals still play a factor in the underutilization of psychological interventions. In addition, it is important to note that there were instances where more than one barrier was chosen, revealing that there are multiple factors at play that keep parents from utilizing psychological

interventions. The qualitative results provide further variety and complexity to the list of barriers identified. In conclusion, there are unmet needs for parents of individuals in the Duchenne Muscular Dystrophy community reflected in the underutilization of and barriers to psychological interventions. Healthcare providers should counsel parents on a variety of interventions as one intervention does not work for everyone.

4.0 Research Significance to Genetic Counseling and Public Health

The aim of this study was to gain an understanding of the parental psychosocial experience within the Duchenne Muscular Dystrophy community. Throughout this study the utilization of psychosocial interventions and barriers to those resources were described. The results have the potential to impact how health care providers, such as genetic counselors and physicians, approach clinical care not only for their patients with DMD, but also for the family as a whole. These results are also impactful to public health because they are informative regarding access to psychosocial resources.

There are three core functions of public health: assessment, policy development, and assurance. Assurance comprises of enforcing laws, ensuring a competent workforce, and evaluating effectiveness. This study can fall under the assurance function.[359] The elements of assurance this study focuses on are linking individuals to the needed psychological health services and evaluating the accessibility of those resources for the DMD community.

The first essential service "[linking] individuals to needed personal health services and [assuring] the provision of health care when otherwise unavailable" correlates to the second aim of this study.[360] It is known that parents of children with DMD are more likely to experience psychological distress than parents who do not have a child with DMD, therefore they should be linked to appropriate psychological interventions[4]. Participants in this study were asked whether they did or did not utilize psychological interventions such as psychotherapy, psychiatry, in-person support groups and online support groups. The results of this study indicate that a majority of participants do not utilize psychological interventions. However, the PHQ-9 screening revealed 94.78% and 91.63% of participants experienced varying degrees of depression symptoms with

42.6% and 23.26% who experienced moderate to severe depression during the diagnosis and disease progression stage, respectively. In addition, GAD-7 results indicated that 94.78% and 93.95% experienced varying degrees of anxiety and 58.26% and 34.41% had moderate to severe anxiety during the diagnosis and disease progression stage, respectively. There is a possibility that parents are not being linked to appropriate health services or that there are other barriers to access psychological resources.

An individual-based health promoting strategy that is relevant to this study and the first essential service includes the principle of multiple methods. The principle of multiple methods is a holistic approach of data collection for formulation of treatment plans and their assessments. It utilizes the biopsychosocial-cultural model which focuses on obtaining multiple perspectives in data collection with both qualitative and quantitative data.[361] Qualitative measures involve the person providing personal, socioeconomic, cultural, and family history information combined with mental health diagnosis guidelines. Quantitative measures utilize screening tools, self-report, and standard measures. Early research showed that mental health screenings were not effective.[362-363] More recent studies show that the mental health screenings have a high degree of validity, feasibility, and clinical utility to use in tracking treatment outcomes and can be useful to screen the general population.[326,327,364-368] Furthermore Duff et al., 2005 found that both patients with cystic fibrosis and their parents approve of screening with a majority of them agreeing that the screening has the capability to accurately label their mental health state.[369] This principle suggests that family-centered health care approaches to treating chronic illnesses should begin in childhood. Any stressor that affects one or more family members, deemed family stress, could affect a family's dynamic, emotional connection, and the overall well-being of the family as a whole.[370] The family-centered approach is to holistically treat not only the patient, but also the family as a

whole.[371] Melnyk et al, 2006 examined the effect of a family-centered approach for parents of children in the neonatal intensive care program and found that those who participated were less stressed than those who did not.[372] Implementing this approach or improving the use in the neuromuscular clinic has the potential to attend to the psychological needs of the parents.

The second essential service "evaluate effectiveness, accessibility, and quality of personal and population-based health services" correlates to the third aim of the study.[360] If participants reported that they did not utilize a psychological intervention, they were asked to select a reason for not utilizing the intervention. This information informs whether there are barriers to access a particular intervention. The results of this study revealed that there are multiple factors that affect access to psychological interventions. For psychotherapy and psychiatry, the top three reasons selected were that participants did not feel that they needed the intervention, time constraints, and financial constraints (Figure 3A). Participants not receiving a referral was the fourth most selected reason for both psychotherapy and psychiatry. The top three reasons selected for in-person and online support group were a lack of support group available, participants did not feel they needed the intervention, and time constraints (Figure 3B). In addition, a qualitative theme that emerged (Table 5) was that there was a lack of resource availability which could be in part due to the pandemic, COVID-19, which occurred as this study was being conducted. During this pandemic access to in person resources was not readily available because of stay-at-home orders and social distancing rules to protect the public. Stopping the spread of the virus became public health officials' main priority. As the threat of the pandemic starts to subside it will be important for public health officials to assure that these psychological interventions become available again. Interventions such as support groups do require an immense time commitment from physicians

and parents, therefore it is important for public health officials to collaborate with all stakeholders to improve access.

Community-based health promotion strategies relevant to this study and the second essential service include the principle of community participation and principle of empowering local people. The principle of community participation states that community members understand their needs the most and are the most qualified to determine what interventions and solutions their community would benefit from. This principle utilizes the asset-based community development model, which focuses on the strengths and capacity at which the community can participate. In turn the community creates policies and activities based on their skill set and capacities.[373] Qualitative data from this model is collected by performing focus groups (members from a community assembled with a moderator to have a discussion around a few questions) which is used to drive social and political change.[374] The principle of empowering local people focuses on empowering communities to take control of factors that have an impact on their mental health and well-being.[375] This principle utilizes community health assessment which empowers the community, ensures knowledge regarding mental health literacy is spread throughout the community, and allows members of the community to actively participate in research.[376] Interventions that can increase mental health literacy include whole community campaigns through social marketing, mental health first aide training, and web-based seminars/campaigns.[377] Furthermore, public health workers and clinicians can gather quantitative and qualitative data that can point out barriers and strengths similar to this study. The results of this study are informative not only to public health departments, but to genetic counselors and other healthcare workers.

As genetic counselors it is an imperative element of the counseling session to address psychosocial elements of the patients and their families as well as provide support resources.[378]

For childhood onset genetic conditions such as DMD not only is the child the patient, but the family as a unit is too. Parents of individuals with Duchenne Muscular Dystrophy experience psychological distress during the diagnosis period[2]. This is the timeframe where genetic counselors are most likely to be involved alongside a neurologist in the neuromuscular clinic. In a clinical sense genetic counselors are uniquely qualified to recognize psychological distress and engage with parents on which psychological intervention would best suit their needs in a non-biased way. It is important to be familiar with anxiety and depression symptoms since a majority of participants in this study experienced some form of anxiety or depression. Genetic counselors could even ask probing questions based on questions from the GAD-7 or PHQ-9 to start a dialogue. When engaging in a discussion, genetic counselors should keep in mind that there are multiple barriers that a parent may have, therefore aiding in problem-solving with the parent could prove to be beneficial. In addition, genetic counseling values interdisciplinary relationships. The results of this study encourage genetic counselors to work with their colleagues to recognize psychological distress, provide credible information on the resources available, implement mental health screening measures, and work with Duchenne Muscular Dystrophy organizations to develop support groups. This could aid in fulfilling the psychological needs of parents of children with Duchenne Muscular Dystrophy and reduce or prevent the burden of acute mental illness.

Appendix A Institutional Review Board Approval

University of Pittsburgh | Institutional Review Board
Office of Research Protections

EXEMPT DETERMINATION

Date:	November 16, 2020
IRB:	STUDY20100066
PI:	Haley Kulas
Title:	Examining parental utilization of and barriers to psychological interventions in the Duchenne Muscular Dystrophy community
Funding:	None

The Institutional Review Board reviewed and determined the above referenced study meets the regulatory requirements for exempt research under 45 CFR 46.104.

Determination Documentation

Determination Date:	11/16/2020
Exempt Category:	(2)(i) Tests, surveys, interviews, or observation (non-identifiable)

Determinations:	
Approved Documents:	• Thesis Survey, Category: Data Collection; • Exempt Application Form, Category: IRB Protocol; • Informed Consent for Survey Script, Category: Recruitment Materials; • Recruitment Email Script, Category: Recruitment Materials; • Recruitment Flyer, Category: Recruitment Materials;

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, Amy Fuhrman.

Please take a moment to complete our Satisfaction Survey as we appreciate your feedback.

Human Research Protection Office 3500 Fifth Avenue, Suite 106 Pittsburgh, PA 15213 www.hrpo.pitt.edu

Appendix B Informed Consent for Survey

Thank you for considering to participate in this survey. This research project is being conducted by Haley Kulas in fulfillment of a master's degree in Genetic Counseling at the University of Pittsburgh.

In this research project, we hope to learn about the experience of parents/guardians of children with Duchenne Muscular Dystrophy (DMD). Specifically, what psychological support resources they have attended or currently attend, including mental health counseling, psychiatry, and support groups. Additionally, we hope to learn of specific barriers that parents/guardians experience, which might prevent access to those psychological support resources. If you decide to participate, you will be asked to complete an online survey containing approximately 40 questions about your mental health experience throughout your child's diagnosis and condition progression, psychological support usage, barriers to those resources, and demographic background. This survey is expected to take 10-15 minutes to complete.

Your participation is entirely voluntary, and you may discontinue at any point. You may decline to answer any question. This survey is anonymous which means that the answers from this survey will not be connected to your name or any other identifying information.

There are no risks to you for your participation in this study, except for the potential emotional distress as you reflect on your experience throughout your child's diagnosis of DMD and the condition's progression. Should you experience any adverse reactions while participating in this survey, you may notify the principal investigator, who can put you in contact with a mental health professional. It is possible that you may not directly benefit by participating in this survey; however, the results from this survey may improve the psychosocial component of genetic counseling of future patients whose child is being diagnosed with DMD.

This study is approved by the University of Pittsburgh's Institutional Review Board. If you have any questions or concerns, you may contact me, Haley Kulas (hmk29@pitt.edu), or my faculty chair, Deanna Steele, MS, LCGC (deanna.steele@chp.edu). Any questions about your rights as a research subject or if you wish to talk to someone other than the research team, please contact the University of Pittsburgh Human Subjects Protection Advocate toll-free at 866-212-2688.

If you know of any parents/guardians who have a child/children diagnosed with DMD that you believe would be willing to complete this online survey, please forward this survey link to them

-Yes, I consent to participate[Begin Survey -No, I decline to participate [Exit Survey]

Appendix C Survey

Thank you for considering to participate in this survey. This research project is being conducted by Haley Kulas in fulfillment of a master's degree in Genetic Counseling at the University of Pittsburgh.

In this research project, we hope to learn about the experience of parents/guardians of children with Duchenne Muscular Dystrophy (DMD). Specifically, what psychological support resources they have attended or currently attend, including mental health counseling, psychiatry, and support groups. Additionally, we hope to learn of specific barriers that parents/guardians experience, which might prevent access to those psychological support resources. If you decide to participate, you will be asked to complete an online survey containing approximately 40 questions about your mental health experience throughout your child's diagnosis and condition progression, psychological support usage, barriers to those resources, and demographic background. This survey is expected to take 10-15 minutes to complete.

Your participation is entirely voluntary, and you may discontinue at any point. You may decline to answer any question. This survey is anonymous which means that the answers from this survey will not be connected to your name or any other identifying information.

There are no risks to you for your participation in this study, except for the potential emotional distress as you reflect on your experience throughout your child's diagnosis of DMD and the condition's progression. Should you experience any adverse reactions while participating in this survey, you may notify the principal investigator, who can put you in contact with a mental health professional. It is possible that you may not directly benefit by participating in this survey; however, the results from this survey may improve the psychosocial component of genetic counseling of future patients whose child is being diagnosed with DMD.

This study is approved by the University of Pittsburgh's Institutional Review Board. If you have any questions or concerns, you may contact me, Haley Kulas (hmk29@pitt.edu), or my faculty chair, Deanna Steele, MS, LCGC (deanna.steele@chp.edu). Any questions about your rights as a research subject or if you wish to talk to someone other than the research team, please contact the University of Pittsburgh Human Subjects Protection Advocate toll-free at 866-212-2688.

If you know of any parents/guardians who have a child/children diagnosed with DMD that you believe would be willing to complete this online survey, please forward this survey link to them

Yes, I consent to participate [Begin Survey]	No, I decline to participate [Exit Survey]

→

List of terms used throughout the survey

Mental Health Counseling-a way to help people with a broad variety of mental health conditions and emotional difficulties involving a talking relationship between therapist and patient. Problems helped by mental health counseling, also called "talk therapy" include difficulties in coping; the impact of trauma, medical illness, or loss of a loved one; and mental health conditions such as anxiety or depression.

Psychiatry-department of medicine that focuses on diagnosing, treating, and preventing mental, emotional, and behavioral conditions. A psychiatrist is a medical doctor who specializes in mental health and assesses both the mental and physical aspects of mental health conditions. Individuals see a psychiatrist for various reasons including panic attacks, feelings of sadness, hopelessness, or anxiousness that never seem to lift, or feeling distorted or out of control. Psychiatrists use numerous treatments such as psychotherapy and medication.

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How many children do you have? Please include both living and deceased children.

- None
- 1
- 2-4
- More than 4

How many children do you have who have been diagnosed with Duchenne Muscular Dystrophy? Please include both living and deceased children.

- None
- 1
- 2
- 3 or more

How many children did you have before having your child who was diagnosed with Duchenne Muscular Dystrophy?

- None
- 1
- 2
- 3 or more

Has your child been diagnosed with Duchenne Muscular Dystrophy within the past 12 months?

- Yes
- No

How old were you when your oldest child with Duchenne Muscular Dystrophy was born?

Under 18 years

18-24 years

25-34 years

35-44 years

45-54 years

Over 55 years

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Before you had your child who was diagnosed with Duchenne Muscular Dystrophy, had you ever been diagnosed with a mental health condition?

Yes

No

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Before you had your child who was diagnosed with Duchenne Muscular Dystrophy, did you ever attend mental health counseling?

Yes

No

Before you had your child who was diagnosed with Duchenne Muscular Dystrophy, did you ever see a psychiatrist?

Yes

No

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Please read each statement and choose 0,1,2 or 3 which indicates how much that statement applies to you **during the time that your child was diagnosed with Duchenne Muscular Dystrophy.**

Do not spend too much time on any one statement. This assessment is not intended to be a diagnosis. If you are concerned in any way, please speak with a qualified health professional.

	0 (Not at all)	1 (Several days per month)	2 (More than half the days per month)	3 (Nearly every day per month)
Feeling nervous, anxious, or on edge	○	○	○	○
Not being able to stop or control worrying	○	○	○	○
Worrying too much about different things	○	○	○	○
Trouble relaxing	○	○	○	○
Being so restless that it is hard to sit still	○	○	○	○
Becoming easily annoyed or irritable	○	○	○	○
Feeling afraid as if something awful might happen	○	○	○	○

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Please Answer the following to the best of your ability

Please read each statement and choose 0,1,2 or 3 which indicates how much that statement applies to you **during the time that your child was diagnosed with Duchenne Muscular Dystrophy.**

Do not spend too much time on any one statement. This assessment is not intended to be a diagnosis. If you are concerned in any way, please speak with a qualified health professional.

	0 (Not at all)	1 (Several days per month)	2 (More than half the days per month)	3 (Nearly every day per month)
Little interest or pleasure in doing things	○	○	○	○
Feeling down, depressed, or hopeless	○	○	○	○
Feeling tired or having little energy	○	○	○	○
Poor appetite or overeating	○	○	○	○
Feeling bad about yourself, feeling that you are a failure, or have let yourself or your family down	○	○	○	○
Trouble concentrating on things, such as reading or watching TV	○	○	○	○
Moving or speaking so slowly that other people could have noticed. Or the opposite- being so fidgety or restless that you have been moving around a lot more than usual	○	○	○	○
Thoughts that you would be better off dead, or of hurting yourself. *National Suicide Prevention Hotline: 1-800-273-8255*	○	○	○	○

If you selected anything other than 0 for the statements in the previous question, how difficult have these problems made it for you to do your work, take care of things at home, or get along with other people?

- Not difficult at all
- Somewhat difficult
- Very difficult
- Extremely difficult
- Not applicable

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During the **time of your child's diagnosis** of Duchenne Muscular Dystrophy did you ever see a mental health counselor?

Yes

No

During the **time of your child's diagnosis** of Duchenne Muscular Dystrophy did you ever see a psychiatrist?

Yes

No

During the **time of your child's diagnosis** of Duchenne Muscular Dystrophy did you ever attend an in-person support group?

Yes

No

During the **time of your child's diagnosis** of Duchenne Muscular Dystrophy did you ever attend an online support group?

Yes

No

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If you did not see a mental health counselor **during the time of your child's diagnosis of DMD**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Never received a referral
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I felt that I did not need to
- Other personal reason

If you did not see a psychiatrist during the **time of your child's diagnosis of DMD**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Never received a referral
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I felt that I did not need to
- Other personal reason

If you did not attend an in-person support group **during the time of your child's diagnosis of DMD**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Lack of local support group available
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I did not feel that I needed to
- Other personal reason

If you did not attend an online support group **during the time of your child's diagnosis of DMD**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Lack of support group available
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I did not feel that I needed to
- Other personal reason

Please Answer the following to the best of your ability

Please read each statement and choose 0,1,2 or 3 which indicates how much that statement applies t
you **as of your child's Duchenne Muscular Dystrophy has progressed over the years**.

Do not spend too much time on any one statement. This assessment is not intended to be a diagnosi
If you are concerned in any way, please speak with a qualified health professional.

	0 (Not at all)	1 (Several days per month)	2 (More than half the days per month)	3 (Nearly every day per month)
Feeling nervous, anxious, or on edge	○	○	○	○
Not being able to stop or control worrying	○	○	○	○
Worrying too much about different things	○	○	○	○
Trouble relaxing	○	○	○	○
Being so restless that it is hard to sit still	○	○	○	○
Becoming easily annoyed or irritable	○	○	○	○
Feeling afraid as if something awful might happen	○	○	○	○

Please Answer the following to the best of your ability

Please read each statement and choose 0,1,2 or 3 which indicates how much that statement applies to you **as your child's Duchenne Muscular Dystrophy has progressed over the years.**

Do not spend too much time on any one statement. This assessment is not intended to be a diagnosis. If you are concerned in any way, please speak with a qualified health professional.

	0 (Not at all)	1 (Several days per month)	2 (More than half the days per month)	3 (Nearly every day per month)
Little interest or pleasure in doing things	○	○	○	○
Feeling down, depressed, or hopeless	○	○	○	○
Feeling tired or having little energy	○	○	○	○
Poor appetite or overeating	○	○	○	○
Feeling bad about yourself, feeling that you are a failure, or have let yourself or your family down	○	○	○	○
Trouble concentrating on things, such as reading or watching TV	○	○	○	○
Moving or speaking so slowly that other people could have noticed. Or the opposite- being so fidgety or restless that you have been moving around a lot more than usual	○	○	○	○
Thoughts that you would be better off dead, or of hurting yourself *National Suicide Prevention Hotline: 1-800-273-8255*	○	○	○	○

If you selected anything other than 0 for the statements in the previous question, how difficult have these problems made it for you to do your work, take care of things at home, or get along with other people?

- Not difficult at all
- Somewhat difficult
- Very difficult
- Extremely difficult
- Not applicable

As your child's Duchenne Muscular Dystrophy has **progressed over the years** did you ever see a mental health counselor?

- Yes
- No

As your child's Duchenne Muscular Dystrophy has **progressed over the years** did you ever see a psychiatrist?

- Yes
- No

As your child's Duchenne Muscular Dystrophy has **progressed over the years** did you ever attend an in-person support group?

Yes

No

As your child's Duchenne Muscular Dystrophy has **progressed over the years** did you ever attend an online support group?

Yes

No

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If you have not seen a psychiatrist, **as your child's Duchenne Muscular Dystrophy has progressed over the years**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Never received a referral
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I felt that I did not need to
- Other personal reason

If you have not seen a mental health counselor, **as your child's Duchenne Muscular Dystrophy has progressed over the years**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Never received a referral
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I felt that I did not need to
- Other personal reason

If you have not attended an in-person support group, **as your child's Duchenne Muscular Dystrophy has progressed over the years**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Lack of local support group available
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I did not feel that I needed to
- Other personal reason

If you have not attended an online support group, **as your child's Duchenne Muscular Dystrophy has progressed over the years**, please select the following reasons that might have prevented you from doing so. (select all that apply)

- Time constraints
- Financial reasons
- Lack of local support group available
- Childcare or caring for sick/disabled loved ones
- Transportation reasons
- I did not feel that I needed to
- Other personal reason

What gender do you identify with?

- Male
- Female
- Non-binary / third gender
- Other
- Prefer not to say

What is your age group?

- 18-24 years
- 25-34 years
- 35-44 years
- 45-54 years
- Over 55 years

What race/ethnicity do you identify with? Choose all that apply

- White
- Black or African American
- American Indian or Alaska Native
- Asian
- Native Hawaiian or Pacific Islander
- Hispanic or Latino
- Native American
- Two or More
- Other/unknown

What is your religious affiliation?

- Protestant
- Catholic
- Mormon
- Jewish
- Buddhist
- Hindu
- Muslim
- No religious affiliation
- Prefer not to say
- Other/ Unknown

What Country do you live in?

If you live in the United States, what state do you live in?

What is your current marital status?

- Single- never married
- Married
- In a domestic partnership
- Divorced
- Divorced and remarried
- Widowed

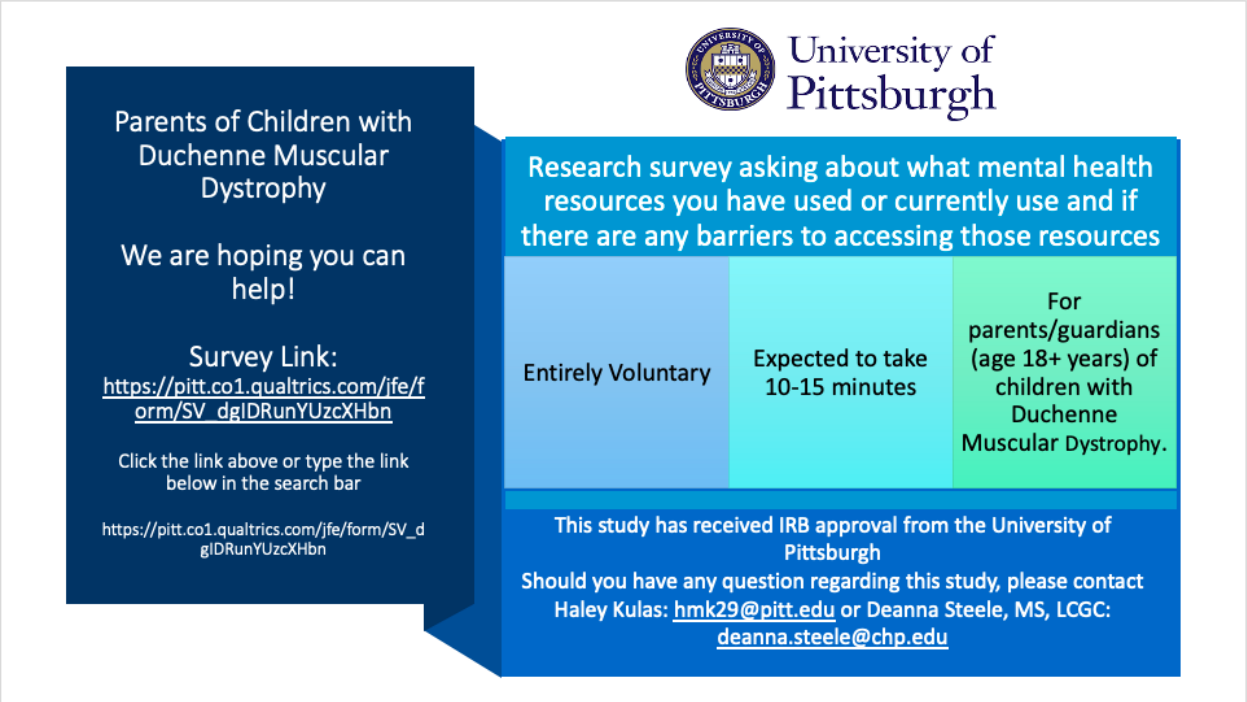
What is the highest level of education you completed?

- High School or less
- Some College
- Bachelor's degree
- Master's degree or higher

What is your current household income in U.S. dollars? This is the income from all the adults who live in your home.

- Less than $20,000
- $20,000-$34,999
- $35,000-$49,999
- $50,000-$74,999
- $75,000-$99,999
- Over $100,000

Appendix D Advertisement for Survey Participation



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